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Editorial Policy

The Journal on Developmental Disabilities is published twice yearly by the Ontario Association on Developmental Disabilities (OADD). The Association has established the following objective for the journal:

> *To foster and promote thoughtful and critical dialogue about relevant issues in the field of developmental disabilities, including those broader social issues that impact on persons with developmental disabilities.*

Content

Each issue features a selection of research, conceptual, informational, and editorial papers. An issue may have a central theme. Reviews of relevant books, movies, websites, software, and other resources are welcomed, as are letters to the editor. In this way we strive to collectively enrich our understanding of issues, encourage stimulating debate among those working in the field, and improve services.

Language

Submissions are invited in either French or English and, if accepted, will be printed in the language of submission with an abstract in the other language. Submissions must be free of any gender and ethnic bias. People-first language (e.g., persons with developmental disabilities) must be used throughout the manuscript (including figures and tables).

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Each submission will undergo a peer review by two or more reviewers (typically from a panel of Associate Editors) with relevant expertise. The authors of the manuscripts will not be identified to the reviewers. Reviewers are asked to judge the manuscript on several criteria including its contribution to increasing our knowledge and clarity of communication. The reviewers will recommend for or against publication and provide their reasons. The reviewers are asked to write in a positive and constructive manner to help the authors improve their work, if necessary.

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Foreward

People with Down syndrome are at risk of developing quite a number of different health concerns that occur more frequently than in the general population. We are very pleased to present in this issue the "Clinicians' Guide to Physical Health Problems of Older Adults with Down Syndrome" that outlines these health concerns. Also, we are honoured to include in this issue an invited editorial "Down Syndrome: The Changing Scene" that highlights increasing advances in our understanding of the characteristics of Down syndrome.

The "Clinicians' Guide" provides an overview of health problems that care providers and professionals should be watching for, particularly in older persons with Down syndrome. Although the title of this article infers that it is primarily for physicians, everyone involved in the care of people with Down syndrome should become familiar with the content. For the benefit of all readers, a glossary of medical and other terms has been compiled from a variety of sources (Appendix I) and appended to the "Guide". A list of "more resources" from local, national and international sources (Appendix II) also has been compiled so that readers may wish to supplement their knowledge and understanding of complications in Down syndrome. For reasons of space, this list of "more resources" is not exhaustive. Readers may be aware of other valuable resources and have suggestions for expanding the glossary. If so, the editorial board asks that these be forwarded to journal@oadd.org so that they can be added to the on-line list.

The authors of "Clinicians' Guide," Dr. Robyn Wallace and Dr. Arthur Dalton, are both eminent clinicians whose work in the field of Down syndrome has had, and continues to have, tremendous impact at the local, national and international levels. Their collaboration in this effort incorporates knowledge gleaned from experience of many different clinicians and researchers, world-wide.

Robyn Wallace is a consultant physician in Internal Medicine who also specializes in tertiary level healthcare for adults with intellectual disability (ID). Half the week, she manages general population inpatient and outpatients at Princess Alexandra Hospital in Brisbane, Australia, and the other half she runs specialized outpatient clinics providing health care for adults with ID at both Mater and Princess Alexandra Hospitals. Among these clinics are specialty clinics for adults with Down syndrome, cerebral palsy,

aging adults, transitional pediatric-adult clinics, and general clinics for all adults with ID. Although these clinics are privately run, she has recently secured funding for clinics to commence in the public health system, an innovation for Queensland. Dr. Wallace's main priorities are clinical, but she completed a Ph.D. where her dissertation focused on *Helicobacter pylori* infection in adults with ID. She has published widely on this topic as well as in other ID areas including physical health issues in aging, Down syndrome, and gastrointestinal conditions. She has given many presentations locally, nationally and internationally. She is also an enthusiastic teacher of medical students. Dr. Wallace was awarded a Churchill Fellowship (specifically the Dorothea Sanders Fellowship in healthcare) on healthcare for adults with ID in 2004.

Arthur Dalton holds a Ph.D. in behavioural psychology. He is Deputy Director of the Center for Aging Studies at the New York State Institute for Basic Research in Developmental Disabilities (IBR/DD) in Staten Island, New York (USA). He has been involved in research on Alzheimer disease for more than 25 years, focusing on the connection with Down syndrome, the development of behavioural and biological markers, and treatment methods. Before moving to New York in 1990, he was based at Surrey Place Centre in Toronto, and appointed to the University of Toronto's Department of Physiology. In Toronto, he pioneered the study of aging and Alzheimer dementia in people with Down syndrome. He established a mobile dementia clinic for adults with Down syndrome, and set a precedent for monitoring individuals longitudinally to check for signs of neurocognitive or neurobehavioural decline, each person serving as his or her own control. Dr. Dalton's ability to develop dementia screening instruments has had great impact in the Alzheimer disease field generally as well as in Down syndrome. Between 1985 and 1991, he was co-principal investigator, along with Dr. Donald McLachlan and Dr. Theo Kruck, of a Surrey Place Centre-University of Toronto phase II clinical trial of desferrioxamine, given by intramuscular injection, in people with moderate Alzheimer disease. Desferrioxamine is an injectable medicine that selectively removes trivalent aluminum and iron ions from the body. This landmark study showed that desferrioxamine slowed the rate of deterioration in daily living skills by a factor of two to three, and reduced the number of deaths, in comparison to treatment with a placebo or no treatment. The impact of this clinical trial continues to gain in momentum as the research community uncovers more and more evidence supporting the involvement of metal ions in Alzheimer disease. In Toronto, Dr. Dalton was the visionary behind a plan to develop an institute dedicated to dementia research. With Dr. McLachlan, his plan took root and bore fruit in the form of the internationally renowned

University of Toronto Tanz Centre for Research in Neurodegenerative Diseases. Based in Toronto, Dr. Dalton also gave much to the community, and is a past-President of the Alzheimer Society of Canada.

Dr. Dalton's impact in the field of intellectual disabilities continued after relocating to the IBR/DD. As well as conducting administrative and research activities at IBR/DD, he has served several years on the board of directors for the National Down Syndrome Congress in the United States. He is the author of numerous journal articles and book chapters and is the co-editor of a seminal text entitled "Aging, Dementia, and Intellectual Disabilities: A Handbook." He is currently the Coordinator of an international, multi-centre, randomized, double-blind, and placebo controlled clinical trial to test the efficacy and safety of Vitamin E that involves 350 adults with Down syndrome, aged 50 years and older. The study is funded by the USA National Institutes of Health.

The author of the invited editorial, Dr. Roy Brown, is an eminent clinician, academic and researcher in the field of Down syndrome. Roy originally worked in the UK as a clinical and educational psychologist before coming to Canada. He was Director of the Vocational Rehabilitation Research Institute in Calgary for many years and was Head of the Educational Psychology Department at the University of Calgary before becoming Dean of Special Education and Psychology at Flinders University, Australia. He is Professor Emeritus at both these Universities. Roy was recognized with the Order of the University of Calgary for his contributions to disabilities nationally and internationally and holds an honorary doctorate from the University of Ghent. Roy continuous to research in the area of quality of life, publishes, and lectures, while also consulting and running workshops. He now lives on Vancouver Island and divides time between consulting, gardening and grandchildren.

We hope that Part 1 of the Supplemental Issue on Down syndrome will be both interesting and useful to our readers.

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Editorial
Down Syndrome: The Changing Scene

Roy I. Brown

Down syndrome has been recognized as a distinct type of disability since it was described by the English physician and educator, John Langdon Haydon Down, as mongolism in 1866. Since then, there have been increasing advances in our understanding of the characteristics of Down syndrome, the most common of the genetic conditions associated with intellectual disability.

There has been a shift in the way we see people with Down syndrome from incompetence to competence, and an increase in our understanding of their health and cognitive abilities. In the 19th and early 20th centuries – indeed until past the middle of the 20th century – people with Down syndrome were all regarded as intellectually disabled, largely falling into the moderate and rather more severe ranges of intellectual disability. Institutionalization for people with Down syndrome was the norm in western society. Now, we understand that people with Down syndrome have a range of ability levels, from severe intellectual disability to low average abilities. Increasingly, we see individuals with average and, occasionally, above average intelligence. For these individuals, the label intellectual disability is increasingly inappropriate, an issue discussed in Brown and Brown (2003). Whatever their intellectual functioning, people with Down syndrome have diverse abilities and this has resulted in most individuals worldwide taking an active role in the societies in which they live. That is not to say all is well and that all challenges are solved.

Changes had small beginnings, and perhaps this is understandable when we recognize that most scientists and society did not believe in cognitive improvements in humans. Until fairly recent times, intelligence was seen as the main measure of human functioning, and was regarded as a constant. Family genetic inheritance was seen as the reason for all kinds of disability and ability. It was believed that human traits ran strongly in families and the possibility that society, as a tool of environment, could cause radical changes, particularly given inherited and genetic structures, was scientifically ignored. The rich were rich and the poor often very poor. There were those born to lead and others to be led. Over time different perceptions were promoted and later became socially accepted, including the fact that

environment could interact with genetic structure to vary the outcome of human development. Interestingly, such views were recognized within botany and animal zoology, but only later in humans.

Development is no longer something restricted to the childhood years, but can continue and needs to be promoted throughout the lifespan. Development is a matter reflected through opportunity as well as motivation. We have come to realize that development is very much dependent on the individual's experiences and the individual's perception of self. Although this in no way reduces the importance of genetics, it does indicate quite clearly how behaviour can change as our environment changes.

Many of the changes that have occurred within the field of Down syndrome have resulted from research in medicine and in the biological sciences. These developments have influenced the way that intervention is carried out, resulting in the correction of a number of the biological defects experienced by children with Down syndrome, thus opening the door to further learning and development. But there are other aspects that are critically important, and to a very large degree follow from these changes in bodily health.

To obtain the full benefits of improvements in health, the individual is dependent on other changes of a societal and educational nature. Early intervention, which has developed into a refined educational art over the past 50 years, is now seen as an important means of providing the necessary stimulation to enable young people with Down syndrome, in many cases, to maintain early developmental stages alongside their peers. But there are other important changes.

In the field of quality of life, which is now playing an increasingly important role in intellectual disability, some groups have been involved more than others. Research in quality of life in the intellectual disabilities field was first promoted with the most vocal and capable of individuals, namely, adults who were young and had mild disabilities; the old and young who were multiply disadvantaged have had to wait in terms of quality of life practice.

In many countries the large institutions have given way to community care, and most children with Down syndrome now live with their families and are subjected to the pluses and the minuses of this development. Families do not always promote effective patterns for their children, let alone children with Down syndrome. Nor do government policies or disability services

necessarily provide the support systems required. One example is the need for respite, frequently seen as a need of the child rather than an urgent and necessary need for family development and stability. This is an issue that requires much further discussion and sensitization of policy.

People with Down syndrome need individual attention, care from parents and brothers and sisters, normal friends, and to not be regarded as different in the local society and therefore excluded. Education, particularly with the advancement of early education, and the inclusion of the child with Down syndrome within the family has led to changes in educational structure so that, in terms of kindergarten and elementary schooling, children with Down syndrome are included within regular groups and classes and have opportunities to learn a similar range of skills to other children. To date, many countries have been less successful in moving the concept of inclusion forward effectively into secondary education (see Forlin, 2005).

Despite this, some individuals with Down syndrome make progress through the secondary cycle of education and a number of these adults now go on to forms of tertiary education including college courses, certificates and diplomas and experience in universities (see Getzel & Wehman, 2005). In the western world a number of individuals with Down syndrome have certificates, diplomas and degrees.

Increasingly, individuals with Down syndrome are moving into a normal environment for work. Yet there still remains, partly because of the blocks within the secondary educational system, a lack of appropriate means of teaching and inclusion. In some places there are signs of giving up and reverting to segregated education. We tend to practice "either/or" practices. There are children who need separate education for periods of time. There are people with Down syndrome who should not be regarded as disabled in educational terms; there are many who can cope with regular education with supports. But there seems to me to be a moral imperative here. Society, through health professionals, has enabled people with Down syndrome to survive early health hazards, consequently they live much longer than they used to. Society, then, surely has a responsibility to pursue and then support in diverse and major ways the progress of these individuals through their lifespan along with their families.

Many of the young adults make their way to part-time and full-time work in unskilled and semi-skilled areas, but counselling of parents and knowledge amongst secondary school personnel may be limited in terms of a) being up to date and b) practice (Grantley et al., 2001). Increasingly, in economically

advanced countries, children with Down syndrome are more frequently welcomed into the family, and are seen as individuals who can learn to function appropriately and bring benefits to their home and family life, and also wider society (Turnbull, Brown, & Turnbull, 2003). Parents who spend intensive time promoting this development often do not find the support they need from community resources. Sometimes this relates to lack of knowledge, lack of application or lack of understanding of legitimate concerns. It is on this front we now must concentrate our efforts.

Lifespan now takes individuals through the mid 50s on average and increasingly into the 60s and 70s. With these developments there are new challenges, just as there are challenges with the increasing number of older people in each of our countries. These challenges in relation to Down syndrome have the same or similar challenges to the rest of the population. But even when similar there are often differences – mental health issues such as depression, dementia, thyroid malfunctioning, along with other challenges such as the issues of an atypical pattern of development leading to puberty, and the correct recognition of premenstrual symptoms, amongst women with Down syndrome (Kyrkou, 2005). The effects of premenstrual syndrome on behaviour are now being documented for the first time, and along with this comes greater understanding of the effects on the woman and on the family.

Dementia is thought to some degree to occur earlier than in the non-disabled population. The symptoms and signs are still frequently overlooked as with hypothyroidism amongst people with Down syndrome despite knowledge being available. One of the challenges is ensuring that this knowledge is readily available and in the right hands: general medical practitioners, nursing personnel and of course, families who are often the people who are first likely to be aware of changes. We need to learn the lessons from the past, recognize that resolution comes at multiple levels, but in a linked and holistic fashion (Schalock et al., 2001).

How are we to do this? This is not just a matter of continuing and reporting ongoing research, but also of applying research to practice and then ensuring general application. Furthermore, the explanations provided by scientists and clinicians need to consider challenges from multiple perspectives. The results often mean we can intervene at a variety of levels and through a variety of interrelated applications.

Health of the individual with Down syndrome is critical, but so are personal experience, social inclusion and opportunities to learn. We tend to talk about

inclusion without sometimes recognizing what exclusion represents. And this becomes more complex now as we reach the older ages, for from the beginning of secondary school onwards through employment, marriage, family life, retirement and the experiences of older age, we still have not critically examined the exclusive behaviours that society, inadvertently or purposely, practices. These are the challenges that now await us.

As usual change is long overdue, since we tend not to deal with issues until problems are well established. There is a need to forecast the challenges in this field. For example, clinicians are seeing more cases of adults with Down syndrome who experience depression. Some clinicians suggest there may be a link with the genetic structure of people with Down syndrome. This may be so, but if an individual is brought up in an inclusive world through school, and then faces an adult world where he or she cannot enter into the normal life style of people around them in terms of friends, partnership, marriage, family, and employment, is there not a possibility that environment is a relevant dimension? Social science, educational policy and practice, along with employment practices, societal views and, of course family action are necessarily involved if we are to deal with such issues. However, if the professionals involved in the child's developing world do not recognize the potential future issues for individuals, catastrophe is likely to occur.

A wide range of professionals are now speaking more meaningfully about the need to provide a broad range of education, both to those who support people with Down syndrome regularly, and to those professionals such as medical practitioners who see them infrequently. Education is also needed to recognize that the one-method approaches that we have often used to date need to be replaced by an understanding that there are "different strokes for different folks." Needs cannot be met if society and its professionals arbitrarily require or demand the same practices for all individuals. Nowhere is this seen more clearly at the present time than in the types of facilities required for people with Down syndrome who are aging. It has been established that inclusion in the community can have a very positive effect on quality of life and living, yet at times such insistence gives rise to poor quality of life. We are fearful of moving back to the old institutional patterns, even though we do not have places of respite of choice that can play a necessary role in meeting individual and family needs in the community.

The changes that I have written about relate in particular to scientific and educational knowledge and the voices that these professionals have in society. Yet probably the motivator for change, and certainly the demonstration that change is possible and effective, was and is to a large

degree advocated and demonstrated by parents, individually and through the numerous societies for people with Down syndrome. Today, there are a wide number of groups made up of parents, people with Down syndrome and professionals (see, for example, the large number of websites of country and state or provincial societies for Down syndrome).

Fundamental scientific research is critical to change. Organizations such as The Down Syndrome Research Foundation in Canada are exploring new avenues of research into brain functioning amongst individuals with Down syndrome. For example, Arlene Young and Larry Roberts are using magnetic (MEG) and electrical (EEG) brain activity to chart auditory brain development in children with Down syndrome (DSRF, 2006). The results should provide a baseline for determining whether acoustic training can enhance auditory developmental trajectories, as it may do in typically developing children.

It is also critical that such research links to application, although such application may not be immediately obvious. A wide range of information from research to practice, however, is now being written for parents and professionals such as the Child and Adult Series of the Down Syndrome Educational Trust (UK). This type of material now needs to be translated into different languages and a few national associations have started this for Down syndrome.

The current issue includes material that is invaluable for practitioners in health and allied fields, and resources relevant to improving the quality of life in every domain for people with Down syndrome of all ages. The challenge now is whether such important contributions will be recognized, read widely, and put into practice in addition to promoting further research.

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Clinicians' Guide to Physical Health Problems of Older Adults With Down Syndrome

Robyn A. Wallace and Arthur J. Dalton

Abstract

Over the last half century, increasing standards of living for the general population and improved healthcare for individuals with Down syndrome (DS) have been coupled with a marked increase in life expectancy of adults with DS. On average adults with DS are reaching at least their fifth or sixth decade of life, though, as they age, they are predisposed to the impact of chronic, or the onset of specific, physical health conditions. Some of these conditions are associated with the syndrome and others with the unique biopsychosocial circumstances facing adults with intellectual disability. The article reviews the common physical health problems of the aging adult with DS from the point of view of the clinician. In doing this, the review addresses each health issue with respect to its prevalence, and its particular diagnostic and management features that, at times, may differ somewhat from the general population. In particular, the article tries to highlight disease prevention strategies and health promotion to promote healthy living into older age for adults with DS.

Introduction

The aim of this review is to provide an overview of diagnostic and management aspects of physical health problems specifically faced by aging adults with Down syndrome (DS). Consideration of the health of older individuals with DS is a relatively new area of public health concern. This is because the average life expectancy for adults with DS in developing countries is now in the mid to high 50s years, and although lower than that of the general population, it has dramatically increased over the last half century when the average life expectancy was below 20 years of age.

Although the adult with DS may not be physiologically prone to premature aging, the literature does suggest a pattern of increase in particular health

conditions in adults with DS as they grow older. By convention, a cut off age of 40 years of age is currently considered the time to start focusing on the "aging health problems" of individuals with DS, though others would suggest that a cut off age of 30 years is more appropriate (personal communication: Dr Helen Beange, Australia). Conceivably, in time, however, this nominal cut-off age of 40 may increase if the life expectancy of adults with DS continues to improve.

The clinician needs to be aware not only of conditions which affect people with DS in particular, but also of other conditions which are the product of lifestyle factors and environmental stressors that predispose any individual with intellectual disability (ID) to certain conditions and result in a different profile of illnesses compared to the age matched general population. Examples of conditions that are unusually common in DS compared to individuals with other forms of ID are vision and hearing problems, autoimmune thyroiditis, diabetes, celiac disease, congenital heart disorders, and dementia of the Alzheimer type. An example of a condition that affects people with ID more frequently than people in the general population is *Helicobacter pylori* infection. Today's older adults with DS already have a somewhat different health profile than their previous generation and their next generation. Examples are the changing prevalences of hepatitis A and B virus infections, and untreated congenital heart disease. However, there is still evidence of disparity in healthcare provision to people with DS and other forms of ID. Adults with DS face the same barriers to good healthcare that most other individuals with ID face, including difficulties in communication, socio-economic disadvantage, insufficient community support, and advocacy. Unfortunately, as well, people involved in the health care of people with DS still are not as knowledgeable as they should be about the risk of comorbid conditions in DS, with the result that adults with DS may be now additionally at risk of the long consequences of long standing undiagnosed pathology in conditions that benefit from early treatment. Examples of conditions that benefit from early treatment are thyroid abnormalities, diabetes, and celiac disease. Furthermore, the diagnostic and management aspects should also be considered in the overall biospychosocial context of the aging patient with DS. That is, the clinician should be consistently aware of the impact of the individual’s varying environmental situations, for example living with aging parents who themselves may be developing age-related conditions, or living in group homes where there may be a turnover of carers. Clinicians should be aware of the consequences of cognitive dysfunction and related behaviours, and how these impact on the obtaining of a history, or on the conducting of an examination or test. The risk of diagnostic overshadowing, the process of

attributing a condition to the disability or behaviour instead of a manifestation of a physical health problem, needs particular attention. Both the patient and their team of caregivers should be intimately involved in the management processes.

The present paper will cover, briefly, the major medical problems in the (1) cardiovascular, (2) respiratory, (3) gastrointestinal, (4) sensory, (5) oral health, (6) skin, (7) neurological, (8) cancer, (9) musculo-skeletal conditions, and (10) endocrine disorders, including aspects that affect (11) health promotion and disease promotion, and (l2) specialized care for aging patients with DS. The clinician should consider reviewing all aspects of function that affect older patients with DS, and the impact of any change of function has on the provision of healthcare. The clinician should be vigilant for all the above conditions in their adult patients with DS, but be particularly watchful for the onset of sensory, endocrine and cognitive problems, the prevalences of which especially increase with age. Where relevant, those conditions where screening is recommended will be highlighted. The paper will conclude with a discussion on the value of specialized clinics for adults with DS.

[Editor's note: Readers should refer to Appendices I and II following this paper for an explanation of unfamiliar terms that are not explained in the text, and for resources that provide more information about specific topics relevant to DS, respectively.]

(1) Cardiovascular Conditions

The three main cardiovascular diseases relevant for older individuals with DS are (a) end stage congenital heart disease (CHD), (b) acquired valvular disease, and (c) coronary artery disease (CAD).

(a) End Stage Congenital Heart Disease Including Eisenmenger's Syndrome and Pulmonary Hypertension

CHD occurs in about half of children born with DS. The most frequent lesions are atrioventricular defect (45%) and ventricular septal defect (35%), but isolated secundum atrial septal defects (8%); isolated persistent patent ductus arteriosus (7%), isolated tetralogy of Fallot (4%), and other lesions (1%) can occur (Tandon & Edwards, 1973). (Refer to Figure 4 of Appendix I for an explanation of normal heart anatomy and function and explanations of the different types of CDH that are common in DS.) Current recommendations suggest routine echocardiograph evaluation of newborns

with DS with appropriate referral for corrective cardiac surgery (Roizen & Patterson, 2003). However, many of today's adults with DS with CHD were not offered corrective surgery years ago, or surgery was not a safe option. Hence, now 20 to 40 years later, these individuals have developed the end stage of CHD, called Eisenmenger's syndrome, and it is this condition that needs consideration in aging adults with DS.

Eisenmenger's syndrome is an incurable condition. It usually is associated with a large ventricular septal defect. The syndrome is defined by the existence of pulmonary hypertension coupled with flow of blood from the right ventricle to the left, bypassing the lungs. Lack of oxygenated blood in the body results in cyanosis. Pulmonary hypertension initially develops because blood shunts through the hole in the septum from the left ventricle to the right one, increasing the volume of blood in the pulmonary vasculature. Damage to the pulmonary vasculature eventually leads to increased pressure in the right ventricle, resulting in a shunting of blood through the septal defect from right to left so that blood cannot enter the pulmonary artery. In addition to the effect of CHD, there is some evidence that a syndromal effect of DS makes the pulmonary arteries more vulnerable to pulmonary vessel damage from the increased pulmonary pressures and subsequent pulmonary hypertension development (Yamaki, Horiuchi, & Sekino, 1983). Once the pulmonary vasculature is damaged enough to result in shunting of blood from the right to the left ventricle, corrective surgery to the cardiac defect does not reverse the pulmonary hypertension and could even hasten death.

The mortality from untreated CHD peaks in young adulthood and middle age and then reduces with age as those with DS surviving to older age do not have untreated CHD (Yang, Rasmussen, & Friedman, 2002). Of all the cardiac-caused deaths, death from untreated CHD and the development of Eisenmenger's syndrome is the greatest frequency cause, though the life expectancy of individuals with DS and treated CHD is likely to be as other individuals with DS without CHD (Baird & Sadovnick, 1987; Mathew, Moodie, Sterba, Murphy, Rosenkranz, & Homa, 1990). In time, as this cohort of individuals with DS and untreated CHD pass on, the prevalence of death from Eisenmenger's syndrome will likely reduce.

Eventually, most patients with Eisenmenger's syndrome develop one or more of the following conditions: (1) symptoms of low systemic output (such as dyspnea, fatigue or syncope), (2) subtle neurologic abnormalities (such as dizziness, headache or visual disturbances) due to erythrocytosis and the resulting hyperviscosity, or (3) symptoms of congestive heart failure

(Vongpatanasin, Brickner, Hillis, & Lange, 1998). Arrhythmias, hemoptysis and cerebrovascular accidents (e.g., stroke) are not uncommon (Bull, Rigby, & Shinebourne, 1985). Physical examination may reveal central cyanosis and clubbing of the nail bed, signs that may be more prominent if systemic vascular resistance falls. The jugular venous pressure may be normal or elevated, with prominent "v" waves. The mainstay medical treatment is essentially palliative including avoidance of medications that have not proven to be beneficial and may cause complications such as hypotension. Antibiotic prophylaxis should be given for any procedure that may cause a bacteremia. Phlebotomy by removal of 500 ml of blood in 30-40 minutes with infusion of an equal volume of isotonic saline or salt free albumin may be used for volume replacement in patients with symptomatic polycythemia or severe erythrocytosis (hematocrit > or = 0.65). Blood pressure should be monitored to avoid hypotension and iron replacement is necessary if iron deficiency develops. Strenuous exercise should be avoided. Surgical options include single or bilateral lung transplantation with closure of cardiac defect, or combined heart-lung transplant, which, in the general population has a 10 year survival rate of about 30%. Unless transplant is an option, at the current time, individuals with DS and untreated CHD who have developed Eisenmenger's will likely die prematurely, and a decision of palliative care measures may be appropriate to institute when symptoms no longer respond to conservative measures.

(b) Acquired Valvular Heart Disease

Adults with DS are at risk for the development of acquired valvular lesions in their early adult years, which may become symptomatic as they age. The common valvular defects found in adults but not children with DS who do not have congenital heart disease include mitral valve prolapse, mitral regurgitation and aortic regurgitation (Romano, Ferrando, Romano, & Pongligione, 1981). The health implications of these types of valve abnormalities include the risk of bacterial endocarditis, the future development of heart failure, and cardiac arrhythmias. Thus, adults with DS should have yearly cardiac history and examinations; if a murmur is audible, they should undergo echocardiography to characterize the type and severity of lesion.

A history of change in exercise performance or syncopal episodes should alert the clinician to more urgent evaluation and cardiology referral. There are already well-developed clinical pathways for advising patients when medical or surgical treatments are warranted. Whenever these patients undergo dental work, antibiotic prophylaxis should be provided.

(c) Coronary Artery Disease

Although coronary artery disease (CAD) is a major cause of mortality and morbidity in the general aging population, this condition appears to be less common among aging adults with DS, though the incidence increases with age (van Allen, Fung, & Jurenka, 1999; Yang, Rasmussen, & Friedman, 2002). Risk factors for the development of CAD include hypertension, diabetes mellitus, hypercholesterolemia, a positive family history, smoking and sedentary lifestyle. There is evidence that rates of hypertension (Morrison, McGrath, Davidson, Brown, Murray, & Lever, 1996; Richards & Enver, 1979), and smoking (Draheim, McCubbin, & Williams, 2002) are lower in the aging population with DS which may somewhat protect these individuals from the development of CAD. Some authors have found lower levels of atheroma in the coronary vessels of older adults with DS at autopsy when compared to the general population (Murdoch, Rodger, Rao, Fletcher, & Dunnigan, 1977), but others have found no difference or higher (adverse) levels of lipid profiles of adults with DS when compared to control groups (Murdoch et al., 1977; Pueschel, Craig, & Haddow, 1992). Moreover, the high rates of sedentary lifestyle, obesity, and elevated fasting insulin observed in this population, could comprise additional risk factors for the development of CAD (Draheim, McCubbin, & Williams, 2002). As the life expectancy of this population increases, an increased trend in CAD may be observed as has been observed in other populations with ID.

Asking patients with DS about chest pain is appropriate for some patients but, for others, a reliable history of chest pain will be difficult to prove. (At least this is the experience of the authors.) An exercise stress test is generally the first line diagnostic test in the general population, but among older individuals with DS, musculoskeletal disabilities or cognitive impairment may preclude this test. Other diagnostic options include a thallium persantin test or a dobutamine stress echocardiograph.

Both of these tests may require preparation of both the hospital staff and patient. Health promotion of the adult patient with DS should include regular surveillance and treatment of lipid profiles, ensuring adequate amounts of exercise, and optimizing body mass index. The management of confirmed CAD requires addressing lifestyle factors, medical therapy such as aspirin and beta blockers and consideration for surgery when appropriate.

(2) Respiratory Conditions

The major respiratory conditions relevant for older individuals with DS include (a) sleep apnea, (b) pneumonia, and (c) pulmonary hypertension

secondary to congenital heart disease. Only the first two conditions will be described in this section. The latter has already been highlighted in section (1).

(a) Sleep Apnea

Sleep apnea (SA) is a respiratory disorder that is expressed as multiple cessations of breathing through sleep that may be due to occlusion of the airway (obstructive sleep apnea (OSA)), absence of respiratory effort (central sleep apnea (CSA)), or a combination of both (mixed sleep apnea (MSA)). The prevalence of sleep disordered breathing among people with DS lies between 31% and 63%, rates higher than in the general population (Lefaivre et al., 1997). Moreover, the prevalence of SA increases and becomes more severe with age among adults with DS (Telakivi, Partinen, Salmi, Leinonen, & Harkonen, 1987). Structural differences in the upper and lower respiratory systems such as smaller mid-facial area, narrowed hypopharynx, macroglossia and alveolar hypoplasia, as well as increasing age-related dysfunction in the respiratory centre in the brain stem comprise factors thought to predispose the aging group with DS to SA (Ferri et al., 1997).

Table 1 highlights some of the clinical presentations of OSA which manifest in cognitive, behavioural and physical health domains. Although links between cognitive dysfunction and SA have been demonstrated (Andreou, Galanopoulou, Gourgoulianis, Karapetsas, & Molyvdas, 2002), it is not clear whether this association is causal or resultant, and it is uncertain what impact treatment would have on cognitive function in adults with DS. The presence of sleep apnea is speculated to comprise an additional risk for pulmonary hypertension in those with untreated congenital heart disease.

Table 1. Clinical presentation of obstructive sleep apnea

Developmental and behavioural problems
Excessive daytime somnolence
Behavioural disturbances
Developmental delay
Failure to thrive
Abnormal sleep patterns
Noisy snoring
Nocturnal insomnia
Gasping respirations
Pauses
Chest retraction
Cyanosis

(continued)

Table 1. (cont'd)

Restless sleep
Enuresis
Worsening of nocturnal seizures
Unusual postures
Long term sequelae
Pulmonary hypertension
Right ventricular hypertrophy (cor pulmonale)
Right sided heart failure
Systemic hypertension
Arrhythmias
Hypoxic encephalopathy including cortical blindness
Polycythemia

(King 1996; Lefaivre et al. 1997)

With a history of suggestive symptoms, the diagnosis of SA requires a sleep study or polysomnography. If SA is confirmed, either central or obstructive, conservative care should include optimization of the person's weight, normalization of thyroid dysfunction, minimization of sedative medications, consideration for oral surgery, or continuous positive airway pressure (CPAP). As with the general population, compliance with the CPAP mask may be low and require formulation of a behavioural and desensitization plan especially in this population. Ideally, formal links with sleep units with a dedicated service for adults with DS could assist in compliance.

(b) Pneumonia

Respiratory infection due to aspiration, or bacterial or viral infections is one of the more common causes of death among aging adults with DS, in contrast to younger individuals with DS (Chaney, Eyman, & Miller 1985; King & Newmann, 1996; van Allen, Fung, & Jurenka, 1999; Yang, Rasmussen, & Friedman, 2002). As few patients with DS smoke, smoking-related lung disease appears to be relatively rare in contrast to the general population. Conditions which predispose the general aging population to pneumonias, such as osteoporosis and secondary reduction of chest wall expansion, appear at earlier ages in the population with DS. High-density living in institutions, lack of exercise, obesity and unfitness, common among populations with DS, likely also comprise risk factors for infectious causes. Given the high rate of aspiration pneumonias, attention should be given to the possibility of gastro-esophageal reflux or swallowing disorder, and management of seizures particularly common in those with reducing cognitive status.

A focus on preventive treatment should include ensuring adequate exercise and fitness, treatment of osteoporosis, avoidance of obesity, vigilance for and treatment of swallowing disorder, as well as yearly influenza and pneumococcal vaccinations every five years.

For active pneumonia, treatment should include antibiotics, with due consideration of whether or not the infection was community or institutionally acquired (the pneumonia acquired in a hospital or another type of institution tends to be far more severe than pneumonia acquired in the community and requires more aggressive treatment), physiotherapy, culture of sputum, incentive spirometry, and supportive care such as attention to hydration, bowels, deep venous thrombosis prophylaxis, and skin care. For pneumonia in patients with advanced (not mild or moderate) dementia, a decision may be reasonably made for a palliative care approach, which includes attention to skin, hygiene, and physical and emotional comfort, rather than intensive medical therapy. This decision should be made in conjunction with the patient's next of kin and medical staff.

(3) Gastrointestinal Conditions

The main gastrointestinal conditions considered relevant for older individuals with DS are (a) celiac disease, (b) Helicobacter pylori (H.pylori) infection, and (c) hepatitis. Like other physical health conditions, some of these appear to be syndrome related, for example, celiac disease, while others, such as H.pylori infection, are related to environmental factors to which people with DS are exposed by nature of having cognitive impairment.

(a) Celiac Disease

Celiac disease, a chronic inflammatory bowel disease induced by an environmental precipitant, gluten, the storage protein of wheat, is associated with symptoms of malabsorption and vitamin deficiencies, fatigue, lassitude, pruritus, osteoporosis, infertility, autoimmune diseases and malignant diseases, especially lymphomas (Green & Jabri, 2003). Previously, celiac disease was thought to affect about 1 in 250 people in the general population. Now, prevalence estimates among Caucasians are as high as 1 in 100 in some studies (Lee & Green, 2006). But among those with DS, its frequency is much higher – up to 17% in one study – but most studies suggest about 7% prevalence (Bonamico, Mariani, Danesi, Crisogianni, Failla, Gemme et al., 2001; Carnicer, Farre, Varea, Vilar, Moreno, & Artigas, 2001). Chromosome 21 carries a gene involved in the regulation of autoimmunity called AIRE (Nagamine et al., 1997; Finnish-German

APECED Consortium, 1997). AIRE is thought to be a transcription factor in cells responsible for the induction and maintenance of immunological tolerance. Because there was no evidence of genetic linkage of celiac disease to chromosome 21 in 21 families multiply affected with celiac disease but lacking DS, Morris et al. (2000) concluded that polymorphic variants of AIRE do not cause celiac disease in DS. However, lack of genetic linkage to chromosome 21 does not rule out the possibility that polymorphic variants of AIRE contribute to the development or expression of celiac disease in DS and in the general population. Furthermore, because the gene product of AIRE is a transcription factor, it is possible that having three copies of AIRE instead of two uniquely predisposes people with DS to a spectrum of autoimmune disorders that affect different glands and tissues, including celiac disease (Sherman & Gagel, 2001).

Table 2. Pathophysiological consequences of malabsorption disorders such as celiac disease

Sign or symptom
Weight loss/malnutrition
Diarrhea
Flatus
Glossitis, cheilosis, stomatitis
Abdominal pain
Bone pain
Weakness
Anemia
Night blindness
Bleeding
Peripheral neuropathy
Dermatitis

Although the typical symptoms of celiac disease include steatorrhea, up to one third of individuals with DS with celiac disease may have an asymptomatic form (Bonamico et al., 2001), hence the importance of screening for this condition. As these studies demonstrating the silence of this pathology have come to light, it is clear that many adult patients with DS seem to have slipped though childhood without being tested for celiac disease. It is important to diagnose and treat the condition because the associated morbidity and mortality can be eliminated by adherence to a gluten free diet. Moreover, as people with DS are already predisposed to nutritional deficiency, and osteoporosis, especially in older age when the impact of these conditions is felt, so identification and treatment of celiac disease may reduce prevalence of these conditions, or at least reduce the conditions' severity.

The formal screening diagnostic process involves serological testing, and if positive, endoscopic biopsy of the small bowel to confirm typical changes, including flattening of the villi and crypt hyperplasia. The diagnostic pitfalls of the tests, both serological and biopsy, are reviewed in Green's recent review (Green & Jabri, 2003). In studies among people with DS, the presence of both anti-tissue transglutaminase and endomysial antibodies appears to correlate very well with positive biopsy findings, though there are some studies to suggest a high false positive rate of anti-tissue transglutaminase antibodies among people with DS (Rumbo, Chirdo, Ben, Saldungaray, & Villalobos, 2002). Total IgA levels should also be measured, as individuals with celiac disease may have reduced levels of IgA leading to falsely negative antibody tests.

The authors suggest the following process for diagnosis and management: Assess the patient with DS for symptoms of celiac disease; screen all adults with DS for celiac disease with IgA anti-tranglutaminase and antiendomysial antibodies and for total IgA. If both antibody tests are positive and IgA is not absent, ideally an endoscopy should be performed to confirm the presence of celiac disease. However, the presence of comorbidities or behavioural problems may make endoscopy a hazardous process. Therefore, if the antibodies are positive and biopsy is not possible, further testing to determine the HLA type is recommended, since the majority of people with celiac disease have a particular combination of HLA antigens. If HLA tests are negative, it is unlikely the patient has celiac disease. If both serological tests are negative, and IgA is deficient, test for IgG antibodies. Once celiac disease is confirmed, strict 100% compliance with a gluten free diet is required.

A gluten free diet seems initially very restrictive with marked social implications. However, given the complete reversibility of the pathology of celiac disease with a 100% strictly gluten free diet, the diet must be imposed. Although the person with DS should be educated and encouraged to participate in the selection of gluten free foods, in most cases it will be the responsibility of the carergivers to ensure 100% compliance and impose the conditions of the diet. Carers and family members must be informed adequately about the diet as if they had the condition themselves. It is prudent in the first six months of treatment to provide supplements of fat soluble vitamins such as A, D, E, K and folate, iron and vitamin B12. At six monthly intervals, serological quantitation could be repeated to assess compliance as these revert to negative with strict compliance. In those who remain symptomatic on a strict diet, consideration should be given to re-endoscope to look for lymphoma or refractory celiac disease.

(b) Helicobacter pylori (H.pylori)

Although adults with DS do not appear to be at any greater risk of acquiring H.pylori infection compared to other adults with ID, many adults with DS are exposed to known biopsychosocial risk factors for H.pylori-related infection and disease (Wallace, Webb, & Schluter, 2002). For example, those with a history of institutionalization have a prevalence of infection up to four times that of the general population (Wallace, Webb, & Schluter, 2002). For those who have never been institutionalized, older age is also positively associated with infection (Wallace, Webb, & Schluter, 2002). Other independent risk factors for infection among the population with ID include having lower skills, greater levels of maladaptive behaviour, or living with flatmates with hypersalivation or fecal incontinence (Wallace, Webb, & Schluter, 2002).

The importance of detection of this infection is particularly relevant for older individuals with ID. The infection is more likely to be acquired during childhood and the longer duration of infection, the greater the risk for peptic ulcer disease or gastric cancer development. Some studies suggest that adults with DS have an increased risk of peptic ulceration and gastric cancer, which, by implication, may be due to H.pylori infection (Hill et al., 2003).

It has been shown that adults with ID do not complain of and their carers do not routinely detect dyspepsia so the symptoms of H.pylori may be silent in this population (Wallace, 2002). Thus, in those with risk factors as outlined, it is important to screen for infection. The choice of screening test includes serology, urea breath test or fecal antigen test. It has been shown that the sensitively and specificity of all these tests among adults with ID are comparable with those results from the general population (Wallace, Webb, Schluter, Forgan-Smith, & Wood, 2003). Only those with greater levels of ability are able to perform the urea breath test, but it has been shown that level of function or behaviour does not affect ability to collect blood or feces as long as there is adequate carer support and motivation. If the patient has alarm symptoms, such as unexplained vomiting, anemia, weight loss, or melena, then an endoscopy should be arranged as is the process in the general population.

Treatment should be given to all patients who test positive, regardless of overt symptoms. Eight weeks after treatment, the individual should be re-tested again, with the fecal antigen test or, if possible, the urea breath test. The family doctor and caregivers should closely monitor the patient for any side effects of treatment. Studies in populations with ID suggest that the eradication rate is somewhat lower than the general population, the side

effects rate higher and the recurrence rate higher, but for the majority of patients, successful eradication is possible and should therefore be offered to any individual who tests positive (Wallace, Webb, & Schluter, 2004). Treatment cures gastritis and peptic ulcer disease, and likely plays a role in ulcer prevention and cancer prevention.

(c) Hepatitis A, B and Autoimmune Hepatitis

In years gone by, children with ID, especially those who were institutionalized, were at high risk for hepatitis A and B infections, compared to the general population. Effective immunization programs are now in place which have greatly reduced the risk of acquiring these infections, and subsequent disease development.

However, older individuals with DS still living may not have had access to hepatitis A or B immunization and therefore may be at risk of acquiring the infections or, in the case of hepatitis B, have had active infection for many years. The risk of liver cancer or cirrhosis development in those who are hepatitis B carriers is highest in those with evidence of high viral replication (positive HepBsAg and HepBeAg). For these individuals, antiviral treatment should be considered in discussion with a specialist gastroenterologist.

All adults with DS regardless of their living situation should be screened for hepatitis A and B immune status. If they are not immune, they should be offered immunization. If the individual has a history of infection with hepatitis A or B, they are considered immune. Likewise their families and carers should be immunized.

A number of autoimmune hepatic disorders occur in older people, including primary biliary cirrhosis, chromic hepatitis and primary sclerosing cholangitis. The latter two conditions have been reported to occur among adults with DS. The data are insufficient to establish whether or not these conditions occur more frequently in people with DS compared to other controls. Nevertheless, these observations support the proposition that people with DS have an increased rate of autoimmune diseases (Kaushik, Kaye, & Clarke, 2000; McCulloch, Ince, & Kendall-Taylor, 1982).

These particular autoimmune conditions are treatable and should be considered in the differential diagnosis along with hepatitis A and B, when evaluating an adult patient with DS who has evidence of impaired liver function, but there is no need to screen for these relatively rare disorders.

(4) Sensory Problems

Vision and hearing impairments associated with aging can affect a wide variety of cognitive abilities, communication skills, independence, and mobility. These are additional disabilities faced by the majority of older adults with DS. Thus, to optimize the health, well-being and function of the older individual with DS, sensory impairments require prompt recognition and treatment.

(a) Vision Impairment

The prevalence of vision impairment is higher across the whole population of people with ID, when compared to the general population, and increases with age and level of disability. Moreover, within the population with ID, the prevalence of vision impairment not only increases with age but is higher in all age groups in those with DS (Kapell, Nightingale, Rodriguez, Lee, Zigman, & Schupf, 1998; van Allen, Fung, & Jurenka, 1999; Van Buggenhout et al., 1999).

Cataracts and acute keratoconus appear to be the main causes of vision loss in people with DS, but strabismus, blepharitis, and high refractive error are also common in the older group with DS and may be more functionally debilitating (Warburg, 2001). Largactil, a commonly misused psychotropic drug among the population with ID, can cause central cataracts and Vigabatrin, an anti-epileptic drug, can cause peripheral vision loss.

In general, the prevalence data suggest that carers and clinicians should assume that vision impairments exist in at least a third of their patients with DS over 40 years, and in every patient with DS aged 65 to 74 years. Formal evaluation by optometrists (for visual acuity) and ophthalmologists (for diagnosis and treatment) should be routinely conducted on a yearly basis for people with DS who are 40 years of age or older. Use of professionals for this assessment is important as it has been noted that carers frequently attribute the additional vision disability to the overall disability syndrome. Corrective surgery, spectacles, environmental modification to reduce the risk of falls and optimize independence, and psychosocial adjustment should all be part of an effective treatment plan, depending on the cause of the vision impairment.

(b) Hearing Impairment

Adults with DS, particularly older adults, are prone to a wide variety of hearing difficulties due to the four types of presbyacusis (sensory, neural,

metabolic and mechanical), infections, and problems with auditory processing. Like vision impairments, the prevalence of hearing impairments among older adults with DS increases with age and is more common in the population with DS compared to populations with other etiologies of ID (van Schrojenstein Lantman-de Valk, et al, 1994).

There is little published research literature on acute ear pathology in older adults with DS. In general terms, for acute ear infection, it is important for the proper management of care to diagnose whether or not the infection is due to otitis media or otitis externa. Giving ear drops to a person who has an acute perforation or has grommets in the ears could potentially worsen the hearing loss due to otoxicity if some of the ear drops pass through a perforation.

Some researchers have found that auditory problems among individuals with DS can interfere with short term memory and language tasks (Marcell & Cohen, 1992), though others have not found this association (Jarrold, Baddeley, & Phillips, 2002). Still other authors have proposed that the presence of abnormal brainstem evoked potentials in people with DS means that central nervous system abnormalities play a role in auditory processing problems (Ferri, Del Gracco, Elia, Musumeci, & Stefanini, 1995).

As in the case with vision problems, formal assessments by audiologists and possibly ear, nose and throat specialists are required to evaluate hearing and diagnose the cause for any impairment, and help develop the management plan (Evenhuis, 1996). The recognition and treatment of hearing impairments or changes in auditory processing is important especially in those with dementia. Treatment may involve the use of hearing aids, treatment of acute ear pathology and/or adjustment in the socioenvironmental conditions to accommodate. Even if only a few people with ID are able to use or regain normal hearing with the use of a hearing aid, adaptation in communication aids may help cope with the loss. The Dutch recommendations for management of hearing impairment includes measures to reduce background noise at home and work, hearing aids, and development of alternative communication aids (Evenhuis, 1996). They recommend that people with DS over 50 years of age should have otoscopy at least twice yearly and hearing assessments every three years. It has been speculated that a portion of hearing impairment could be prevented if everyone with DS had been regularly examined for ear problems beginning at a younger age.

(5) Oral Health

Congenital anomalies in the cranial morphology, particularly the jaw and the dentition may contribute to the high prevalence of oral health problems

specific for people with DS (Hennequin, Faulks, Veyrune, & Bourdiol, 1999; Lomholt, Russell, Stoltze, & Kjaer, 2002). There is some evidence that parotid salivary gland flow rate is slower in people with DS compared to the general population and particularly among older people with DS compared to younger people (Chaushu, Becker, Chaushu, & Shapira, 2002; Chaushu, Yefenof, Becker, Shapira, & Chaushu, 2002). This could potentially adversely affect oral hygiene, and also increase the risk for pneumonias. Interproximal bone loss may also be higher among people with DS (Gabre, Martinsson, & Gahnberg, 2001) as part of generalized osteoporosis, also compromising dentition. Some environmental factors may also contribute to poor dentition either by inadequate support for oral hygiene or nutritional deficiencies. Systemic diseases which occur more commonly in people with DS, such as celiac disease, may also affect tooth structure.

Hennequin and other authors (Hennequin, Allison, & Veyrune, 2000; Hennequin, Faulks, Veyrune, & Bourdiol, 1999) recommend that dental visits with emphasis on preventive activities should be scheduled three to four times per year. The aim of the preventive measures should be to prevent the onset of dental caries, especially on the occlusal surfaces, and to minimize the progression of periodontal disease. The desired level of oral hygiene can rarely be obtained by means of traditional mechanical plaque control and all published papers reinforce the need for regular dental review, flossing and antiplaque measures (e.g., clorhexidine). Carers of adults with DS should be motivated and aware of the importance of taking an active role in providing oral hygiene.

The need for anesthesia for dental assessment and treatment in uncooperative patients, those with atlanto-occipital instability and other comorbid conditions including polypharmacy, and the need for antibiotic prophylaxis in those with Eisenmenger's disease and valvular heart disease, constitute issues that require special consideration for dental work in people with DS. A published case report by Schmidt and colleagues (Schmidt, Wolter, Lenschow, & Kienast, 2001) provides more details. For dental procedures that cause gingival bleeding and oral surgery, 2.0 grams of amoxicillin administered orally is required 1 hour before the procedure. If the patient cannot take medication orally, then 2.0 grams of ampicillin should be administered intramuscularly or intravenously 30 minutes before the procedure (or the oral regimen for penicillin allergic patients, namely Clindamycin, 600 milligrams, administered before the procedure).

(6) Skin

There is little in the literature on the specific dermal problems in older people with DS. Among children with DS, frequently documented conditions are palmoplantar hyperkeratosis, xerosis, seborrheic dermatitis, fissured tongue, geographic tongue, and cutis marmorata (Ercis, Balci, & Atakan, 1996). Lip lesions including angular stomatitis and fissures are particularly common among adults with DS, although there is no evidence that these lesions increase with age (Scully, van Bruggen, Diz Dios, Casal, Porter, & Davison, 2002). Usual attention to hygiene, avoidance of detergents and use of sorbelene is recommended as baseline care.

Pressure ulcers are a particular concern in all elderly patients and presumably also those with DS, particularly if they have Alzheimer disease. Seiler and colleague (Seiller & Stahlin, 2000) underscored the important practical care issues. These include attention to pressure ulcer risk factors from neurological or physical conditions that diminish or prevent involuntary bodily movements, hypotension, hypoxia, anemia, infections, fever, malnutrition, and impaired skin health. The classic ulcer sites are the skin over bony prominences: sacrum, heels, trochanteric, lateral malleolus, and ischial bony prominences. The best treatment is prevention, which includes carefully managing the magnitude of interface pressure by a static prevention method (e.g., with specialized mattresses, pillows), managing the duration of interface pressure by frequently turning the patient (preferably every two hours), and by treating or eliminating any other risk factors. Once the ulcer is present, the principles of good clinical practice include complete pressure relief, debridement of necrotic tissue, and treatment of local infection by systemic antibiotics, wet wound dressings, and the elimination of risk factors.

(7) Neurological Conditions

(a) Dementia and Alzheimer Disease

Individuals with DS are uniquely vulnerable to Alzheimer disease (AD) with many showing clinical signs of dementia after the age of 50 years. The signs and symptoms of AD in persons with DS are similar to those that occur in AD in the general population, though they may be more difficult to discern and they appear much earlier in life. The average age of onset of dementia in persons with DS has been reported to be 52.8 years (Janicki & Dalton, 2000). Dementia of the Alzheimer type in DS sometimes is called "DAT".

The increasing life expectancy for persons with DS, with average age at death estimated at 55.8 years (Janicki et al., 1999), is stimulating a growing interest and concern over diagnostic issues and the development of appropriate interventions in the management of their health problems, notably AD.

Prevalence studies of dementia in aging adults with intellectual disabilities and/or DS are relatively few. Sixteen studies, all but one involving fewer than 200 research participants, conducted over the period from 1980-1996, have been critically reviewed (Holland, 1998). A state-wide, informant survey of persons with DS yielded a prevalence estimate of dementia among 22% of adults 40 years of age and older (Janicki & Dalton, 2000). A cross-sectional population-based study of individuals with DS 30 years of age and older from one health district, with a population of 280,000, found estimated age-specific incidence rates of all dementias ranging from 22.2 % to 26.3%. All were conducted independently without sharing of data (Holland et al., 2000). The instruments being used to establish the presence/absence of dementia differed from study to study, and most were not supported by post-mortem neuropathological diagnoses.

The problems of diagnosis and assessment of dementia have long been recognized (Aylward et al., 1997; Burt & Aylward, 1998; Oliver, 1998; Holland, 1998). The ICD 10 criteria are useful for diagnosing dementia of the Alzheimer type in DS. These are shown in Table 3.

Table 3. ICD-10 criteria for dementia and Alzheimer disease (Aylward, et al., 1997)

1. Decline in memory. Most evident in the learning of new information, although the recall of new information, although the recall of previously learned information may also be affected in more severe cases. The impairment applied to both verbal and nonverbal material.
2. Decline in other cognitive abilities. Characterized by deterioration in judgment and thinking, such as planning and organizing, and in the general processing of information. Deterioration from a previously higher level of performance should be established.
3. Awareness of the environment. Absence of clouding of consciousness for a period of time sufficiently long to allow the unequivocal demonstration of decline in memory and other cognitive functions.
4. Decline in emotional control or motivation, or a change in social behaviour. Changes are manifested in at least one of the following: (a) emotional lability, (2) irritability, (3) apathy, (4) coarsening of social behaviour.

(continued)

Table 3. (cont'd.)

5. Duration. Decline of memory and other cognitive functions must be present for at least 6 months.

1. All criteria for dementia are met.

2. Exclusionary criteria. No evidence from the history, physical examination, or special investigations, for any other possible cause of dementia, a systemic disorder, or alcohol or drug abuse.

3. Onset and progression. For a diagnosis of Alzheimer disease, there must be evidence of gradual onset and continuing cognitive decline.

In spite of the high degree of variability in the nature, severity, extent, and age of onset of the signs of dementia, for practical purposes, the progression from early to late stage disease has been characterized to some degree, as shown in Table 4.

Table 4. Staging, diagnosis, care, and time frame for persons with Down syndrome suffering from dementia of the Alzheimer type (DAT) (Dalton and Janicki, 1998)

Early stage	
Behaviour:	Memory, orientation, communication, work, and social skills dysfunction
Diagnosis:	Suspicion of DAT
Care:	Support compensatory activities
Time frame:	1 to less than 5 years
Middle Stage	
Behaviour:	Pronounced losses and declines in language, comprehension, orientation, short-term memory, daily living skills, personality along with confusion, withdrawal
Diagnosis:	Probable diagnosis of DAT
Care:	More intense supervision, controls for wandering and safety, modification of daytime activities
Time frame:	2-5 years
Late Stage	
Behaviour:	Almost complete loss of all higher functions, memory, mobility, social, personality, and mobility, frequent incontinence, seizures
Diagnosis:	Probable Alzheimer disease (A diagnosis of definite AD requires histopathological evidence of disease)
Care:	Complete supervision required; frequent nursing care, and appropriate medication to deal with infections, ulcers, etc.
Time frame:	1-2 years

The published literature has focused mostly on changes in cognitive functions (Dalton et al., 1999; Devenny et al., 2000; Kay et al, 2003; Holland et al., 2000: Temple et al., 2001), adaptive and daily living skills (Prasher, 1998), and movement disorders (Dalton & Fedor, 1998; Evenhuis, 1998) as indicators of dementia of the Alzheimer type. Many co-morbid conditions (Dalton & Janicki, 1998), including sensory impairments (Evenhuis, 1998), other neurological conditions (Wherrett, 1998), depression (Burt, 1998) and other psychiatric disorders (Thorpe, 1998) may complicate dementia diagnosis. A full physical health history, examination and appropriate investigations, assessment of mental health, review of the environmental and social situation should be provided for all adults with DS with functional decline to assess the possible causes, as the differential diagnosis of dementia is considerable; but even in the presence of dementia, patients' physical, mental, environmental and social conditions should be regularly assessed and managed to optimal state.

The differential diagnosis of functional decline in persons with DS has been described succinctly by Pary (1992). He identifies the following disorders which are common in persons with DS: depression, hypothyroidism, infection, folate or B12 deficiency, hearing impairment, visual impairment, malignancy, joint problems of the neck, knee or hip, AD and sleep apnea. The authors would suggest that, in addition, all of the physical health conditions mentioned in this paper need to be considered. Disorders with no apparent predilection for persons with DS include subdural hematoma, brain tumours, and normal pressure hydrocephalus. The workup (Pary, 1992) for functional decline in DS should include: sleep and weight graphs, vital signs, complete blood count, screening blood chemistries, electrolytes, thyroid function tests, folate and B_{12} levels, urinalysis, electrocardiogram, chest X-ray, brain imaging study, neuropsychological testing, assessment of daily living skills, graph of work productivity, and consideration of audiological, ophthalmologic, orthopaedic and sleep laboratory consultations.

The standardization of neuropsychological tests, behaviour rating scales, and related instruments for use in the functional evaluation of persons with DS at risk for AD have been plagued by persistent problems, with no single test or battery having been identified that has generally acceptable psychometric properties (Aylward et al., 1997). Longitudinal studies are few (see review by Prasher, 1998) and almost none are supported by post-mortem neuropathological diagnoses of AD. Many rating scales have been developed to assist in diagnosis and assessment (Dalton, et al., 2002; Deb & Braganza, 1999; Gedye, 1995; Kay et al., 2003) and many pre-existing tools intended for other purposes have been modified for use with persons with intellectual

disabilities. Aylward and her colleagues (Aylward et al., 1997) have reviewed the usefulness of more than 15 rating scales and neuropsychological tests to assist in diagnosis of dementia and AD in persons with intellectual disabilities using ICD-10 World Health Organization criteria. Ball and colleagues (Ball et al., 2004) recently reported new data on the usefulness, validity, and reliability of the modified CAMDEX informant interview as a tool for use in the diagnosis of dementia in adults with DS. Prasher (2004) has recently published a validated office-assessment of dementia among adults with DS, *The Adaptive Behaviour Dementia Questionnaire* (ABDQ). The value of this tool is that it can be administered by a clinician via a brief five minute interview with the caregiver, while in their clinical setting and without the need for special training.

Today, there are five treatments approved by the USA Food and Drug Administration (FDA) for AD in the general population, namely, tacrine (Cognex), donepezil (Aricept), rivastigmine (Exelon) and galantamine (Reminyl) – based on their properties as cholinesterase inhibitors – and memantine. Donepezil has received the most attention in persons with DS (Lott, et al., 2002; Prasher, et al., 2002).

(b) Epilepsy

There are distinct peaks to the incidence of seizures among people with DS which result from different causes (Stafstrom, 1993). In the perinatal period seizures are commonly a consequence of medical complications and infection. Then, in the first year, infantile spasms account for a significant percentage of seizures. After that, new onset seizures are uncommon until the teenage years when there is an increase in incidence probably due to fall-related head injuries. This is followed by a distinct increase in seizure frequency that occurs in the fourth and fifth decades of life, coincident with the onset of Alzheimer's-type brain changes.

Some factors specific to DS engender enhanced seizure susceptibility accompanying the onset of AD. Electroencephalograph (EEG) changes that occur in older individuals with DS also appear to precede the onset of any deterioration or dementia (Tangye, 1979). The frequency of new onset epilepsy in patients with AD who do not have DS is about 10%, whereas in individuals with DS and AD, the frequency exceeds 75% (Evenhuis, 1990; Lai & Williams, 1989). Other possible causes of seizures to consider in older people with DS include subdural hematoma acquired through trauma or cerebral abscess which has been reported as an excessive cause of death (Hill et al., 2003).

In general, seizures are associated with an increase in mortality and cognitive decline. Antiepileptic medications may also produce adverse behavioural and cognitive side effects. Moreover, older adults with DS may also be at increased susceptibility to physical trauma and fractures from seizures as they may have pre-existing osteoporosis and mobility imbalance. The onset of seizures can also create stress and anxiety for caregivers, particularly if coping with the onset of dementia at the same time.

Thus, new onset seizures in adults with DS warrants, as in any patients with seizures, a strategic approach to diagnosis and management of the seizure syndrome, such as that proposed and published by the IAASID working group (IASSID, 2001), and one which involves and supports the caregivers. This requires involving the carers to give a witness description of the events described as seizures, EEG recording, cerebral imaging, and documentation of response to treatment (IASSID, 2001). After the diagnosis of the seizure syndrome, seizure syndrome-specific antiepileptic medication should be commenced. Although an aim is to eliminate seizures, the clinician and team should work closely together to monitor any cognitive and behavioural impacts of antiepileptic medication and balance the outcomes. In a series of five patients with DS, dementia and epilepsy, phenytoin was observed to result in adverse events, which were initially attributed to the dementing process. Sedation, unsteadiness and abrupt cognitive deterioration were reversed when the phenytoin was ceased. The authors (Tsouris, Patti, Tipu, & Raguthu, 2002) postulated that the side effects were due to the increased sensitivity of the remaining neurons to the known side effects of phenytoin. They recommended against the use of phenytoin for seizure control among people with DS and dementia-associated epilepsy.

(8) Cancer

The prevalence of all-cause cancers appears to be lower among adults with DS compared to the general population (Hasle, 2001; Yang, Rasmussen, & Friedman, 2002), and the profile different compared to aged-matched peers in the general population. The overall lower prevalence of all cancers may be related to the lower life expectancy among adults with DS, environmental or protective chromosomal factors. As in the general ID population, as the life expectancy has increased, so has the incidence and prevalence of cancers (Jancar, 1990) and this may be seen in future data among those with DS. Whereas lifestyle factors such as reduced smoking rates among the population with DS may protect against cancer, other lifestyle factors, such as institutional living (which is associated with increased rates of hepatitis B and H.pylori infections, both of which are associated with cancers), and genetic causes related to trisomy 21 could promote the risk of cancer.

Hill and colleagues (Hill et al., 2003) found adults with DS had an increased risk of liver cancer and elevated mortality due to stomach (speculated association with H.pylori infection), liver (speculated associated with hepatitis B infection), and gallbladder cancers, although the risk estimates were based only on a few observations. Leukemia was common in children but was not common among older adults with DS. An excess prevalence of testicular cancer was reported among males with DS (Satge, Sasco, Cure, Leduc, Sommelet, & Vekemans, 1997), a not surprising finding considering the higher prevalence of cryptorchidism in this population. However, there is some evidence also that chromosome 21 gene expression is directly associated with a predisposition for testicular cancer. There was also an increased risk of penile and brain cancer reported in that study. The risk of breast cancer was found to be lower among women with DS in one study (Satge, Sasco, Pujol & Rethore, 2001). The data on cancer prevalence among older individuals are minimal and tend to be based on small numbers.

Preventive measures such as yearly clinical examination of testes and surgical referral in cases of undescended testes should be routine. Those at risk should be screened for H.pylori and hepatitis B. In addition, without evidence to the contrary, the authors suggest that the same strategies for cancer screening in the general population apply for the population with DS. Women with DS who are older than 50 years should undergo bi-annual mammogram testing, (as it is the recommendation for the general population); stool guaic test over the age of 50; cancer check up (including thyroid, prostate, lymph nodes, oral region, and skin) should be considered for anyone with DS over the age of 50 years. These strategies may be modified if more data become available.

As part of good clinical management, people with DS should be offered standard therapy for cancers, chemotherapy, surgery and radiation or combination, which are adapted for the specific care setting. Treatment should take into account the cognitive impairment in explanation, comorbidities and environmental factors such as level of support needed. Both the patient with DS and their caregivers must be informed about the treatments and expected side effects in appropriate language.

The response to cancer treatment in older people is limited not by age itself, but by the presence of comorbidities, choice of multidisciplinary treatments, choice of treatment modality, physical and psychosocial supports that are accessible. Cancer treatments are often prolonged and involve many hospital visits. The regular caregivers should always accompany the patient with DS for the hospital visits, and may be required to play an advocacy role. Taking good records at the home is important to provide to the hospital based

medical team. For adults with DS who have advanced dementia, a palliative approach may be considered the most appropriate management strategy.

(9) Musculoskeletal Conditions

The major musculoskeletal issues relevant for older individuals with DS include (a) osteoarthritis of the neck and hip, (b) osteoporosis and (c) carpal tunnel syndrome.

(a) Osteoarthritis

In the general population, the risk factors for osteoarthritis include increased age, female sex, genetic factors, obesity, major joint trauma, occupation, hypermobility, and conditions such as diabetes, hyperuricemia and hypertension. For the hip joint, osteoporosis appears to be a protective factor. Hyperuricemia, hypermobility, obesity, diabetes and osteoporosis are frequently present among the aging population of adults with DS.

The development of cervical spine abnormalities is of particular relevance among older individuals with DS, although these abnormalities differ from those in younger people with DS. Widening of the atlanto-axial occipital joint is characteristic of children with DS, but in adults, a predominance of degenerative joint disease is more common (Maclachlan et al., 1993). This may lead to spinal cord stenosis and the development of upper motor neuron signs and symptoms. The patient with significant cervical spine abnormalities causing spinal cord compression may present with difficulty with gait, stiffness or weakness on one side or the other or both, affecting the arms as well as the legs, but without involvement of the cranial nerves.

In a study by Olive and colleagues (Olive, Whitecloud, & Bennett, 1988), among 105 adults with DS aged between 21-50 years, by the age 40 years, 70% of participants had lower cervical spine spondylosis; hypereflexia was seen in 34%, clonus in 17%, and ataxia in 17% of participants. In Maclachlan's study (Maclachlan et al., 1993), degenerative cervical spine disease was seen in 64% of people with DS aged between 21 and 60 years of age, and 29% of controls. Severe degenerative cervical spine disease was recorded in 19% of cases and 26% showed moderate severity disease.

Appropriate investigations include a plain film of the neck, magnetic resonance imaging (MRI) or computerized tomography (CT) and myelography when there are neurological signs. Decompressive surgery

may be required, and must be undertaken with due consideration of co-existing comorbidities and planning for rehabilitation.

Hip disease occurs in 8%-22% of people with DS (Cristofaro, Donovan, & Cristofaro, 1986; Hresko, Mccarthy, & Goldberg 1993; Shaw & Beals, 1992). There is an increased prevalence of dislocation, dysplasia, slipped epyhypisis, Perthe's disease, avascular necrosis and osteoarthritis (Shaw & Beals, 1992). Abnormalities in the anatomy of the hip in people with DS include an increase in acetabular depth, increased capsular laxity and an increased range of movement appear to make the hip joint more vulnerable to the development of arthritis (Kioschos, Shaw, & Beals, 1999). Increasing life expectancy for people with DS may mean that debilitating hip disease becomes more common.

Several small studies report good outcomes from hip surgery in adults with DS and severe hip disease (Kioschos, Shaw, & Beals, 1999). Preoperative evaluation of a patient with DS should take into account other orthopedic considerations including patella hypermobility, instability of the upper cervical spine, as well as other comorbid conditions such as cardiac anomalies, and possible predisposition to infections. No infection has been reported in any of the studies on hip arthroplasty (Kioschos, Shaw, & Beals, 1999). Careful postoperative monitoring is necessary to detect infection of the urinary tract and deep venous thrombosis because patients with DS may not report symptoms. Post operatively, the patient should have access to rehabilitation facilities and a specified program of exercise, which caregivers help provide.

(b) Osteoporosis, Falls and Fractures

Everyone loses bone mass with age, resulting in age-associated increases in skeletal fragility.

Table 5. World Health Organization (WHO) criteria for osteoporosis

Normal	BMD within 1 standard deviation of young adults reference mean
Low bone mass (osteopenia)	BMD between 1.0 and 2.5 standard deviations below young adults reference mean
Osteoporosis	BMD 2.5 standard deviations or more below the young adult reference mean
Severe (established) osteoporosis	Osteoporosis as above with one or more fragility fractures

BMD = bone mineral density

Osteoporosis is more common in the population with ID compared to the general population (Center, Beange, & McElduff, 1998; Kioschos, Shaw, & Beals, 1999; van Allen, Fung, & Jurenka, 1999). Bone mineral density is lower in children and adults with DS compared to the general population (Kao, Chen, Wang, & Yeh, 1992). Lower levels of exercise among young people with DS, hypogonadism in men and earlier menopause in women (Schupf et al., 1997), lower level of calcium, increased conditions of malabsorption (e.g., from celiac disease), institutional living, lack of exposure to sunlight, anticonvulsant medication, thyroid abnormalities, lower levels of exercise as adults, and impaired mobility, may all contribute. The physician and carer should take a proactive stance to look for and treat these secondary (treatable) causes in care of the younger individual with DS.

The risk of fractures is increased in people with osteoporosis. People with DS who have gait abnormalities, who are taking antipsychotic medications, who have epilepsy, who have sensory problems, and who show evidence of declining cognitive function conceivably have a particularly heightened risk for falling and sustaining a fracture. These same factors are applicable for the general aging population when assessing risk of falls. Aging factors, per se, may also contribute to the risk of falls and trauma by their association with arrhythmias, poor neuromuscular condition, muscular weakness, defective posture, hearing and vision impairments, unsuitable environment, confusion and malaise.

Prevention is one of the key management strategies for osteoporosis and fractures. Weight bearing exercise and adequate calcium intake are important, particularly during youth. In the general elderly population, exercise involving balance, hip protectors, reduction of antipsychotic medication and benzodiazepines, and falls assessment at the residence, are important preventive and management strategies. Assessment and correction of secondary causes form a vital part of the effective care management.

Medications to minimize further reduction in bone density include combined calcium (at least 1.2-1.5 grams per day) and vitamin D supplementation, hormone replacement therapy and bisphosphonates. The decision to use female hormone replacement therapy for females needs to take into account family history of breast cancer and the necessity to perform mammograms every two years to deal with the risk of cancer from this intervention, and also cardiac implications. The use of testosterone for men must take the risk of medication-produced elevations in blood pressure and cholesterol. Continuing testicular and prostate examinations are advised in this context. Bisphosphonates administered once weekly are effective in

reducing fractures in the general elderly population. The response of people with DS to these treatment interventions is unknown. A baseline documentation of bone mineral density should be performed before treatment is started. Documentation of efficacy should be a guiding principle whenever a patient is exposed to a potentially toxic drug. This same principle should be applied in the treatment of osteoporosis (May, 1999).

(c) Carpal Tunnel Syndrome

Carpal tunnel syndrome denotes a compression neuropathy of the median nerve as it passes beneath the flexor retinaculum at the wrist. The symptoms include progressive hand weakness or clumsiness due to impairment of the finer movements of the hand. It is associated with attacks of pain, tingling, and numbness of the affected hand with the little fingers unaffected. Characteristically the attacks are nocturnal.

Patients with DS have been found to have a high prevalence of carpal tunnel syndrome (Christensen, Peter, Nielsen, & Mai, 1998). In an uncontrolled study of 48 patients with DS, 16 of whom were aged between 30 and 50 years, 38% were found to have carpal tunnel syndrome and an additional 19% had probable carpal tunnel syndrome – a percentage that was marginally higher than in the younger study participants. Only two patients had complaints or clinical signs suggesting carpal tunnel before the examination. The authors speculated whether or not the presence of hypothyroidism or amyloid deposition in the brain may contribute to these findings. Based on this study, the physician should examine the hands for signs of wasting and take a history of loss of use of hands or distress at night with painful hands.

(10) Endocrine Disorders

The major endocrine disorders that manifest in older adults with DS include (a) thyroid dysfunction, (b) diabetes mellitus and (c) hypogonadism. Hypogonadism is not reviewed here specifically, but mentioned in relation to other conditions such as osteoporosis, and cancer risk.

(a) Thyroid dysfunction

Most of the prevalence studies conducted among adults with DS over the age of 40 years show a prevalence of thyroid dysfunction of at least 30%, with a higher incidence with age and in females with DS. The cause of

thyroid dysfunction in adults with DS is thought to have an autoimmune basis in the majority of cases (Percy et al., 1990). Hypothyroidism is more common than hyperthyroidism in the population with DS.

Primary hypothyroidism (due to thyroid organ dysfunction) is characterized by elevated thyroid stimulating hormone (TSH) and low levels of thyroxine (T4 and/or T3). When the level of TSH is above the reference range and the level of T4 below the range, this combination is called overt hypothyroidism and therapy is indicated. The condition where TSH is elevated and the T4 is still in the normal range is called subclinical hypothyroidism, and may still be associated with clinical features so that replacement therapy needs to be considered on an individual basis. Conversely, when the TSH is low and the T4 is high, this condition is called overt hyperthyroidism, and when TSH is low with normal T4, this is called subclinical hyperthyroidism. Secondary hypothyroidism (due to pituitary/hypothalamic disorders) can de difficult to diagnose. The TSH can be low, normal or, paradoxically, slightly elevated. Free T4 is relatively low but usually only at the lower limit of normal. Other evidence of pituitary or hypothalamic disturbances should be present, particularly secondary hypogonadism.

The clinical diagnosis of hypothyroidism may be difficult in people with DS as some of the features of hypothyroidism are also features of DS or aging in general. In his review, Prasher (Prasher, 1999) highlighted the poor correlation between clinical assessment of thyroid dysfunction and biochemical results. He also emphasized the need to beware of over diagnosis of thyroid dysfunction (Prasher & Haque, 2005).

Table 6. Common symptoms and signs of hypothyroidism in the elderly

Common symptoms of hypothyroidism in the elderly
Fatigue, weakness, lack of energy, excessive sleepiness
Intolerance to cold
Anorexia, muscle cramps, parasthesias
Dry skin, coarse skin
Constipation
Hoarseness of the voice
Decreased memory, mental slowing
Swelling of the hands, face or extremities

Untreated thyroid disease can be associated with reversible cognitive impairment, making it an important issue for the general elderly population

as well as for those with DS (Thase, 1982). However, it does not seem to be associated with the presence of established dementia or depressive symptomatology (Prasher, 1995) although Percy and her colleagues (Percy et al., 1990) provided some data suggesting a connection to Alzheimer disease.

Due to the absence of long-term follow-up studies of thyroid disorders, a high index of suspicion is required and a recommendation has been made for screening (Prasher, 1999):

- Normal thyroid status: repeat tests every two years
- Definite hypothyroidism: immediate thyroxine replacement (check response no earlier than 6 weeks later)
- Subclinical hypothyroidism: repeat tests every year and check thyroid autoantibodies
- Definite hyperthyroidism: consider for beta blockade, radioactive therapy or medication
- Previous treatment: monitor yearly

Laboratory confirmed hypo- or hyperthyroidism should be evaluated for causes and then treated with replacement thyroxine or anti-thyroid medications, respectively. An Endocrinologist or Internal Medicine Physician is recommended to be involved at the initiation of therapy. Follow-up tests of thyroid levels in response to therapy are generally not useful at earlier than 8-12 week intervals.

(b) Diabetes Mellitus

The exact prevalence of diabetes mellitus among adults with DS is uncertain, though it is probably more common among people with DS compared to the aged matched general population. A Scottish survey in the mid-90s reported that the prevalence of type I diabetes (insulin dependent) among adults with DS was between 1.4% and 10.6% (Anwar, Walker, & Frier, 1998), a rate which was apparently considerably higher than that in the general population of that study environment, and increased with age. In contrast, a study in 1968 reported that the prevalence of diabetes among adults with DS aged over 20 years of age was not different to that of the general aged matched United States population (Milunsky & Neurath, 1968).

The complications of diabetes include retinopathy, neuropathy, nephropathy, and peripheral and coronary vessel disease. One study found that the prevalence of retinopathy and proteinuria was much lower among adults

with DS when compared to a comparable study group from the general population (Fulcher et al., 1998). The possible protective factors included lower blood pressure, myopia (suggested to be protective against diabetic retinopathy), abnormalities in growth hormone secretion or responsiveness in patients with DS.

Lifestyle factors important in the treatment of diabetes, include a low glycemic factor diet, weight control, smoking abstinence, and exercise. In addition, insulin or oral hypoglycaemic agents may be required to control the blood glucose. Maintenance of the blood pressure below 120/80 reduces the risk of future complications as does tight control of the blood glucose levels. A side effect of tight glucose control, however, is hypoglycemia which is a dangerous medical condition especially as the older patient with DS may not be able to report this symptom. This risk of silent hypoglycemia has led some endocrinologists (McElduff, 2002) to advocate the maintenance of a higher blood sugar level than would normally be aimed for in the general population. This strategy is also employed by geriatricians in caring for their older patients. The addition of an angiotension converting enzyme inhibitor and lipid lowering therapy should be considered, but the risk of hypotension and falls or muscle side effects of the lipid lowering agents must be considered. Urinary protein or microalbumin should be evaluated every year. The eye review could be performed annually in people with DS because they are at risk for other eye diseases in addition to those caused by diabetes.

Patients themselves should be encouraged to play a role in management of their diabetic disease. People with DS and their carers should receive suitable diabetic education. In situations where there is a high turnover of carers, education of carers should be regularly repeated. There may be special techniques to assist in the self-management of diabetes for people with cognitive impairment (Moore, Ewell, Geffken, Johnson, & Silverstein, 1995).

(11) Health Promotion and Disease Prevention

Health promotion can be considered as any action taken to maximize physical and mental health, and well-being among populations and individuals. Disease prevention, a related concept, includes interventions that occur before the initial onset of a disorder to prevent the development of the disorder. Health promotion and disease prevention strategies can be applied across the lifespan, and one should never be considered too old or young to address these. Among older individuals attention to such strategies may minimize the impact of “normal aging” and age-related diseases.

Table 7. Framework of management of healthcare delivery

Biopsychosocial approach to aging issues

- Includes physiological, psychological, social, functional, environmental, health and disease, assessment of change from baseline

Knowledge of baseline function, health and social

- Level of function (e.g. Adaptive Behaviour Scale I) as younger to middle aged adult
- Level of behaviour (e.g. Adaptive Behaviour Scale II) as younger to middle aged adult
- Health profile, diary
- Family history, residential history
- Social networks
- Life story

Knowledge of the scope of the health issues in older adults with Down syndrome

- Cardiovascular (Valvular heart disease, Eisenmenger's syndrome, congenital heart disease, coronary artery disease risk factors, ability to exercise, endocarditis risk)
- Respiratory (Sleep apnea, recurrent pneumonia, pulmonary hypertension; oral health)
- Sensory (Vision, hearing)
- Gastrointestinal (Bowel function, hepatitis, H.pylori risk assessment, celiac disease)
- Endocrine (Thyroid disorder, diabetes, female and male hormone status, testicular examination)
- Musculoskeletal (Osteoporosis, arthritis, carpel tunnel, falls, gait stability, home assessment for falls, exercise)
- Health promotion (Dental, skin, exercise, nutrition, immunization, mammograms, prostate)
- Medication review (avoidance of polypharmacy, medications to increase risk of falls)
- Dementia (Diagnosis, treatment, epilepsy)
- Anesthetic risk
- End of life discussion and palliative care

Logistics of health care practice

- Regular care, regular family doctor
- Be organized – regular appointments
- Regular GP – aware of unusual presentations
- Longer appointments
- Informed carer for appointments
- Ask about aging health checks

(continued)

Table 7. (cont'd)

• Become informed of healthy life for aging people and incorporate into lifestyle • Not necessarily adopt a palliative approach • Adaptations for investigations without compromise • Home-hospital interface – have documentation for hospital staff • Consideration given to specialized health services for adults with Down syndrome
Commitment to ensuring the same quality of care and access to services for aging adults with Down syndrome as other aging population
Knowledge of the legal system and consent issues for that locality

(a) Weight and Nutrition

Eating disorders, such as anorexia nervosa, have been reported among people with DS, but by far, obesity is the main weight problem. Many studies have confirmed that obesity and not just overweight is common along adults with DS. In a study by Bell and colleagues (Bell & Bhate, 1992), approximately 71% of adult men and 96% of women with DS compared to 50% of men and 63% of women with other causes of intellectual disability were categorized as overweight or obese. These findings are in marked contrast to reports of obesity in 40% of men and 32% of women from the general population. Obesity is a risk factor for sleep apnea, coronary artery disease, hypertension, diabetes mellitus, osteoarthritis, impaired mobility and complicates physical examination and treatments of older patients.

There is a growing appreciation of some of the relationships between diet, exercise, disability status, and degree of social integration with body mass index even after the effects of diet, exercise and physical status variables are partitioned out of the analyses. For example, lifestyle variables such as friendships, social opportunity and physical competency are potent predicators of body mass index (Fujiura, Fitzsimons, Marks, & Chicoine, 1997). In the management of obesity, social factors associated with eating habits and healthy lifestyle are critical factors.

The responsibility to address obesity should be tackled at the community, and individual levels. People with DS should be encouraged to participate in healthy eating choices, but where their cognitive impairment prohibits this carers should also be well informed of appropriate nutritional guidelines and

impose these. In the group home setting where there is a rotation of carers, effective management of diet for residents may be more complex and formal assistance from dieticians should be encouraged. So called "junk food" should not regularly be used as a reward. Appropriate caloric restriction and exercise should be incorporated in the management of obesity.

Older adults with DS should also be assessed for particular nutritional deficiencies. Vitamin D deficiency is particularly common among those who have a history of institutionalization. Oral vitamin D replacement is available. Calcium should be provided through low or reduced fat or skim milk, yoghurt or cheese. Folate, iron and vitamins A, D, E, K, B12 may be deficient in those with untreated celiac disease and should be replaced as part of the initial treatment of this condition.

(b) Exercise

Published reports suggest that cardiovascular fitness, even after training, in people with DS may be poorer than in the general population, and in people with other causes of intellectual disability (Climstein, Pitetti, Barrett, & Campbell, 1993; Fernhall et al., 1996; Millar, Fernhall, & Burkett, 1993). Unfortunately, none of these papers take into account medical comorbidities as confounders in the interpretation of their results. Others have shown that special programs increase the fitness of adults with DS (Heller, Hsieh, & Rimmer, 2002).

For older people in particular, regular exercise has been associated with a reduction in falls, improvements in aerobic capacity, muscle strength, flexibility, physical performance, prevention of falls, improvements of sleep quality, improvements in mood and well-being, improvements in psychomotor skill, reduction of anxiety and depression, increased longevity, and decreased risk of cardiovascular disease (Lamb, 2000). For older individuals it is recommended that people undergo a simple assessment of baseline fitness, including postural stability and risk of cardiovascular complications. In addition, exercises should be simple, instructions should be provided to patients and carers and time for practice should be made available. Regular follow ups and reinforcement of exercise principles, progression of prescription to maintain training intensity and minimize boredom should be routinely implemented.

Exercise is an important part of healthy lifestyle, but the level should be supervised and adjusted for the presence of mobility, cardiac, and neurological impairments. For example, those with congenital heart disease should not exercise strenuously but light exercise is not contraindicated.

(c) Mobility and Falls Prevention

Older individuals in the general population and those with DS who have impaired mobility, sensory and cognitive problems, polypharmacy and risk factors for osteoporosis, are at high risk for fractures. To reduce this risk the person in their home environment should undergo a falls assessment, usually by a community-based occupational therapist, to limit the hidden obstacles and risk of injury. Wearing hip protectors and rationalization of medications may also reduce the risk of fractures.

(d) Immunization

Immunizations against influenza yearly and every five years for the pneumococcus are recommended for aging adults with DS. Hepatitis A and B immunizations have probably become standard practice in many countries as a result of advances in knowledge about the conditions. At least a third of older patients have probably had hepatitis A infection; individuals with natural immunity do not require immunization. Assessment of hepatitis A IgG status will clarify the status (a positive IgG implies past infection and no need for immunization). Tetanus injections should also be given to individuals over 50 years of age.

(e) Planning for Anesthesia in Older Adults With DS

Patients with genetic disorders with or without multiple congenital anomalies present unique challenges to the healthcare provider responsible for administering sedation and anesthesia during surgical or other technical procedures. Patients with DS have special health-related needs requiring attention before successful sedation or anesthesia is given, such as cervical spine and cardiac considerations. In addition, the uncooperative patient can be a challenge and special attention must be given to make the patient feel as comfortable as possible. Special behaviour assessments may be necessary before sedation or anesthesia administration to meet or gain the trust of the patients and family to alleviate problems. Desensitization and allaying anxiety are important for successful sedation of patients with special needs (Butler, Hayes, Hathaway, & Begleiter, 2000).

In general, the greater the number of comorbidities, the greater the risk associated with sedation or anesthesia (Krauss & Green, 2000). There may also be oro-pharyngeal or other physical factors which affect ability to provide sedation or anesthesia. Among people with DS, the particular inherent risks associated with general anesthesia are well documented. They

include hypotonia, flat facies, cardiac defects, cervical spine arthritis or instability, short neck, obesity, intestinal, and rib defects. It is recommended to evaluate vertebrae and rib anomalies radiologically, and to evaluate cardiac, intestinal and endocrine disorders among patients with DS before performing anesthesia (Butler, Hayes, Hathaway, & Begleiter, 2000). People with DS may also have an abnormal sympathetic nervous system, which may be the result of neuroleptic drugs and not necessarily an inherent feature of the trisomic condition. Case studies describing the type of care required have been published (for example (Meitzner& Skurnowicz, 2005).

Antibiotic prophylaxis should not be forgotten for invasive procedures among those with congenital heart disease. Attention to respiratory toilet is important post operatively, particularly in those with a risk of sleep apnea. General anesthesia is possible in adults with DS and Eisenmenger's syndrome but it may require a multidisciplinary approach (Bozich & Albert, 1990).

Specific protocols have been developed for use with pediatric patients with disabilities who require sedation or anesthesia. These protocols include attendance by a specialist nurse, pre- and post- procedure observations, and application of established standards for discharge. These protocols have proven to be very safe. The development of a sedation/anesthesia protocol for adults with DS is advocated. Such a protocol might include a pre-operative checklist of possible comorbidities that the anaesthetic physician would review with the patient and their carer, a physical examination and possibly some other investigations. A trial of desensitization might be appropriate in some cases. Staff involved in the protocol would be informed and prepared for the special communication and physical needs of people with intellectual disability. Such a protocol developed for adults with ID requiring sedation for EEGs has been shown to be safe and effective (Wallace, 2003).

(12) Specialized Care for Aging Patients With DS

Specialized DS clinics have been established in many centres for children and adults. Published reports have described the enormous success of the multidisciplinary approach in the effective management of the needs of patients with DS (Chicoine, McGuire, Hebein, & Gilly, 1994; 1995; Lovell & Saul, 1999).

A healthcare service for people with DS should include first a mechanism for providing comprehensive assessments of aging adults with DS run by clinicians with knowledge and expertise, and secondly, a consultancy service for other health professionals dealing with healthcare problems that

occur in people with DS. The logistics of delivering quality healthcare include the involvement of a local family doctor who is known to the patient, the involvement of a carer who is very familiar to the patient and who can take along the medications and healthcare plans and medical notes from the residential house. Longer time needs to be allocated for medical appointments. The presence of an advocate (professional carer, relative or friend) is needed. Case management should be competent and comprehensive and actively involve the patient in decision making. Health care strategies should be communicated to the rest of the household workers. The healthcare worker should build up a list of reliable and professional local facilities, allied healthcare workers for healthcare provision, vision assessments, cooperation with the local hospital and development of sedation anesthesia protocols.

Patients with DS who have a terminal illness (and their carers) need facilities and support services to cope with illness. Palliative care professionals should undergo training with respect to issues of care of adults with ID. Such professionals should be able to tell the patients and carers what is going to happen now, and give individuals the time to absorb the information. Primary and palliative care workers have an intimate knowledge of the physical needs and the expected course of the disease, while carers have an intimate knowledge of the patient. Working together and sharing their expertise will ensure that physical and emotional distress are adequately evaluated and treated (Tuffrey Wijne, 2002). The person with ID often has a very firm and important place in his or her social environment. The impending death will mean a profound loss and a massive change in family or household dynamics. This loss must be recognized, respected and supported.

Specialized services are not always available, so the team assisting the patient with DS must do their best to access generic health services. For patients with DS who present to hospital it is important that well known carers stay with the patient to provide comfort, to assist in history to medical staff, to highlight the new symptoms and response to treatments, and to implement and participate in discharge planning and follow up. Pre-prepared documentation for medical staff describing the patient as a person may assist medical staff, who often feel intimidated in dealing with adults with intellectual disability. The carer may also play the role of an advocate, ensuring optimal care, informing medical authorities of names of the appropriate legal authorities for decision making and consent. Informing the medical staff about the patient's baseline cognitive skills, sensory impairments and usual demeanour and personality are also important in assisting medical staff to interact and assess response to therapy.

Finally, the optimal provision of healthcare of the aging adult with DS needs to be placed within their overall careplan for health and well-being. This means addressing issues of finances and family trusts, residential situations now and for the future, adequacy of level of care, end of life decisions, maintaining networks of friends and families, post retirement activities, and dealing with grief due to loss of other family members, such as parents. Acquiring an accurate documentation of baseline function and cognition during younger adult years is extremely important. Table 7 outlines a framework by which to view the health and well-being of aging adults with DS.

In conclusion, the optimal healthcare and well-being for older adults with DS requires an organized approach in which the clinician is knowledgeable about the specific health conditions and can adapt the generic diagnostic and management processes to suit the individual with DS in their particular setting.

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Appendix I

Special Terms and Explanations Relevant to the Health of Individuals with Down Syndrome

Except where noted, explanations of standard terms provided below have been assembled from a number of different sources. Readers are encouraged to explore the sources listed at the end of the glossary and others for more information about unfamiliar terms.

Acute. This means that a condition develops suddenly.

Alveolar hypoplasia. Alveoli are a crucial part of the respiratory system. They are the air sacs of the lung in which oxygen from the air enters the lungs and carbon dioxide from the body is released to the air. They resemble bunches of grapes attached to the ends of the bronchioles. The respiratory system is sometimes called the respiratory tree. The larynx flows into the trachea, which is the tree trunk. The trachea divides into two tree limbs, the right and left bronchi. Each bronchus branches off into multiple smaller bronchi, branches that are distributed throughout the lung tissue. Each bronchus divides into tubes of smaller and smaller diameter, finally ending in the terminal bronchioles. The term hypoplasia refers to underdevelopment of a tissue or organ. The term alveolar hypoplasia means that the lung weighs less than normal and there are fewer alveoli than normal.

Alzheimer disease. A progressive disease of the brain characterized by loss of nerve cells. It affects many areas of cognitive function including memory, language, behaviour, and daily living skills. It is the most common form of dementia. Characteristic features of the brain in people with Alzheimer disease are amyloid plaques and neurofibrillary tangles. Amyloid also accumulates in the blood vessels of the brain. See behavior symptoms for changes in typical behavior that are characteristic of different stages of Alzheimer disease. Less that 10% of cases of Alzheimer disease are caused by mutations in the amyloid precursor protein (APP) gene on chromosome 21, or the presenilin 1 and presenilin 2 genes on chromosomes 14 and 1, respectively. Most cases are thought to result from a combination of genetic predisposing factors, environmental factors and/or metabolic factors. A healthy lifestyle, ongoing education, regular physical activity, and cholesterol control, may play a role in prevention of Alzheimer disease. Individuals with Down syndrome are thought to develop Alzheimer-like dementia

20-30 years earlier than the general population because most have three complete copies of chromosome 21 instead of the normal two (Prasher & Percy, 2003, pp. 793-807.)

Amyloid beta protein. See beta-amyloid (β-amyloid).

Amyloid plaques. Areas of degeneration in the brain associated with deposits of a protein called beta-amyloid (β-amyloid), a breakdown product of a larger protein called amyloid precursor protein (APP). Amyloid plaques come in two forms: diffuse and neuritic. The diffuse plaques are devoid of structure. The neuritic plaques appear more organized and contain processes of nerve cells called neurites. It is thought that the generation of β-amyloid from APP leads to a type of immune reaction, with the formation of debris and inflammation around the central amyloid core. Because amyloid will bind the metal ions zinc, copper and iron, some researchers believe that amyloid plaques play a role in defending the brain against metal ion toxicity. (Prasher & Percy, 2003, pp. 793-807.)

Anemia. A deficiency of red blood cells and/or hemoglobin. Hemoglobin is the protein in cells that carries oxygen. Lack of hemoglobin results in lack of oxygen in tissues and organs. There are several causes of anemia. These include: iron deficiency, folic acid deficiency, certain viral illnesses, blood loss, exposure to certain toxins such as lead, and certain genetic conditions. The main symptoms are tiredness or weakness. Other symptoms can include pale skin, fast heartbeat, shortness of breath, chest pain, dizziness, cognitive problems, numbness or coldness in the extremities, and headache.

Antibiotic prophylaxis. Treatment with antibiotics to prevent bacterial infection.

Aortic regurgitation. The backflow of blood from the aorta into the left ventricle of the heart, owing to malfunction of the aortic valve. The aortic valve is between the heart's left ventricle and the aorta, the large artery that receives blood from the heart's left ventricle and distributes it to the body. Regurgitation means the valve doesn't close properly, and blood can leak backward through it. This means the left ventricle must pump more blood than normal, and will gradually get bigger because of the extra workload. Aortic regurgitation can range from mild to severe. Some people may have no symptoms for years. But as the condition worsens, symptoms will appear. These can include fatigue (especially during times of increased activity), shortness of breath, edema (retention of fluid) in certain parts of the body such as the ankles, heart

arrhythmias (abnormal heartbeats), and angina pectoris (chest pain or discomfort caused by reduced blood supply to the heart muscle). See "Heart" for details about normal heart function.

Aphasia. A condition characterized by either partial or total loss of the ability to communicate verbally or using written words. A person with aphasia may have difficulty speaking, reading, writing, recognizing the names of objects, or understanding what other people have said. Aphasia is caused by a brain injury, as may occur during a traumatic accident or when the brain is deprived of oxygen during a stroke. It may also be caused by a brain tumour, a disease such as Alzheimer's, or a brain infection, like encephalitis. Aphasia may be temporary or permanent. Aphasia does not include speech impediments caused by loss of muscle control.

APGAR score. This was devised in 1952 by Virginia Apgar as a simple and repeatable method to quickly and briefly assess the health of newborn children immediately after childbirth. The Apgar score is determined using five simple criteria. Each of the five criteria is given a value from zero to two. The sum of the five values is the APGAR score, which can range from zero to 10. The five criteria for the score are: skin colour, heart rate, reflex irritability (the level of newborn irritation in response to stimuli such as a mild pinch), muscle tone, and respiration.

Arrhythmias. Abnormal heart rhythms.

Arteriosclerosis. See Atheroma.

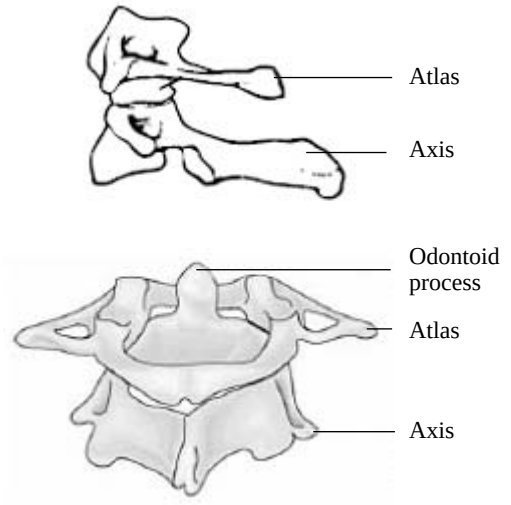
Aspiration. The term can refer to inhaling purposefully (such as breathing in oxygen or inhalants) or inhaling accidentally (such as sucking food into the airway). It may also refer to procedures that involve the use of suction for removal.

Atheroma. An atheroma (plural, atheromata) is an abnormal fatty deposit which develops within the walls of arteries over time. Veins do not develop atheromata, unless surgically moved to function as an artery. The disease resulting from atheromata is called atherosclerosis. The term arteriosclerosis is sometimes used interchangeably with atherosclerosis, which is not correct, as the former term refers to any condition in which the arteries become harder and less flexible

Atlanto-axial instability. This term refers to an increased flexibility between the first and second (atlas and axis) bones (vertebrae) of the neck. Since the vertebrae surround and protect the spinal cord,

instability of the joint could place the spinal cord at risk for injury. The second cervical vertebra, the axis, has a finger-like extension on its upper and front surface called the odontoid process. A transverse ligament holds the odontoid process against the front inner surface of the atlas, or the first cervical vertebra above it. Presumably, as the result of laxity of this ligament in Down syndrome, the odontoid process is able to rock and/or slide backwards when the neck moves, especially when the head flexes forwards. The odontoid process in this circumstance impinges to some extent on the spinal canal that lies directly behind it and therefore on the spinal cord and on the various nerve roots that emerge from it in this area.

Figure 1. Lateral and posterior views of the articulated atlas and axis



Corel Gallery 2, 1995, by permission.

Atresia. Absence of, or closure of, a normal body opening or passage.

Atrial septal defect (ASD). This is a term encompassing several defects in the atrial septum that allow blood flow between the left and right atria. See "Heart" for information about normal heart function.

Atrioventricular canal (AV canal). This is an old term for atrioventricular septal defect (see below).

Atrioventricular septal defect (AVSD). A complex heart problem that involves several abnormalities of structures inside the heart, including an atrial septal defect, a ventricular septal defect, and improperly formed mitral and/or tricuspid valves; in AVSD there is a lack of separation of the four chambers of the heart. AVSD is sometimes called "endocardial cushion defect". During the first 8 weeks of pregnancy, a structure called the endocardial cushion does not fuse with the septum, resulting in this defect. See "Heart" for information about normal heart function.

Atrium. One of the two upper chambers of the heart. See "Heart" for information about normal heart function.

Audiologist. A professional who has received special training in evaluation of hearing, on the effects of hearing on communication, and rehabilitation of hearing loss.

Auditory. Of, or having to do with, hearing or the organs of hearing. See "ear" for information about how we hear.

Auditory processing (of language). The ability to understand spoken language. Information moves through the neurological system more slowly for children with auditory processing disorders. Sometimes it takes much longer for them to understand what has been said.

Autoimmune disorders. Conditions in which a person's immune system attacks the body's own cells, causing tissue destruction. Examples of autoimmune disorders that are common in Down syndrome are autoimmune thyroiditis, diabetes (type I or insulin dependent) and celiac disease.

Autoimmune hepatitis. A disorder in which the body's immune system attacks cells of the liver causing it to become inflamed.

Autoimmune thyroiditis. Inflammation of the thyroid. See "Thyroid" for a diagram of where the thyroid is located in the neck and an explanation about thyroid function.

Bacteremia. Bacterial infection. Some surgical and dental procedures cause a brief bacteremia. Bacteremia is common after many invasive procedures, but only certain bacteria commonly cause bacterial endocarditis.

Bacterial endocarditis. An infection of the heart's inner lining (endocardium) or the heart valves. This results when certain types of bacteria in the bloodstream lodge on abnormal heart valves or already damaged heart tissue.

Behavioral symptoms. Changes in typical behaviour that are markers or indicators for a disease or psychiatric pathology. As an example, behaviors associated with the three main stages of Alzheimer disease are given below:

> *Stage I - Early* (duration 1-3 years): memory loss (particularly short-term memory; changes in personality; difficulties with language; disorientated in time; lost in familiar places; loss of motivation; signs of depression and aggression; and loss of interest.
> *Stage II - Middle* (duration 2-5 years): very forgetful; loss of self care skills; greater dependency on others; increased speech difficulties; wandering; seizures; reduced mobility; behavioral problems; and hallucinations.
> *Stage III - Late* (duration 4-8 years): marked intellectual deterioration; inability to recognize family, friends, carers; dependence on others for dressing, washing, feeding, toileting; immobility; bladder and bowel incontinence; more severe seizures; limb rigidity and flexed postures; marked physical deterioration (Prasher & Percy, 2003, p. 799.)

Beta-amyloid protein (β-amyloid protein). A breakdown product of a large protein encoded by a single gene located on chromosome 21, called the amyloid precursor protein (APP). It is thought that there is a problem with APP metabolism in all persons who develop Alzheimer disease. Most individuals with Down syndrome have 3 copies of the APP gene instead of the normal two. It is thought that this predisposes individuals with Down syndrome to developing Alzheimer disease at a much earlier age than people in the general population (Brown & Percy, 2003, pp. 793-807, by permission).

Beta blockers. Medicines that affect the body's response to certain neurotransmitters. This class of drugs was first developed for the treatment of certain heart conditions and high blood pressure. Later, they were also found to be useful in glaucoma, migraine, and some psychiatric disorders such as performance anxiety, tremors secondary to lithium, and movement disorders caused by some drugs used in the treatment of psychosis.

Biliary cirrhosis. A liver disease that slowly destroys the bile ducts in the liver. Bile, a substance that helps digest fat, leaves the liver through bile ducts. When the ducts are damaged, bile builds up in the liver and damages liver tissue. Over time, the disease can cause cirrhosis and may make the liver stop working. The term cirrhosis means an irreversible scarring of the liver.

Body mass index (BMI). This is defined as the ratio of a person's weight (in kilograms) over the square of the height (in metres). In adults, the BMI is an indicator of health risks. The higher the BMI, the greater the risk of developing health problems (e.g., type 2 diabetes). Someone with a BMI of 26 to 27 is about 20 percent overweight, which is generally believed to carry moderate health risks. A BMI of 30 and higher is considered obese.

Bowel. Another name for the intestine. The small bowel and the large bowel are the small intestine and large intestine, respectively.

Brain stem. This is the stalk of the brain below the cerebral hemispheres. It is the major route for communication between the forebrain, the spinal cord, and peripheral nerves. It controls various autonomic functions (functions that we are not normally in control of) such as respiration and the regulation of heart rhythms as well as perceptual functions such as the primary aspects of sound localization.

Brain stem [auditory] evoked potentials. Evoked potentials are tests to measure the electrical impulses produced by the nerves of the central nervous system in response to a stimulus supplied by the tester, hence "evoked." Brain stem auditory evoked potentials test nerves involved in hearing by using sounds to stimulate and evaluate lesions in the brain stem.

Canthus. One of the two corners of the eye. The canthus nearest the eye is the inner or nasal one; that nearest the side of the head is the outer or temporal one.

Cataract. A clouding of the natural lens, the part of the eye responsible for focussing light and producing clear, sharp images. The lens is contained in a capsule. As old cells die they become trapped within the capsule. Over time, the cells accumulate causing the lens to cloud, making images look blurred or fuzzy. For most people, cataracts are a natural result of aging. See "Eye" for a diagram of parts of the normal eye.

Celiac disease. A disease of the digestive system in which the inside lining of the small intestine (mucosa) is damaged after eating wheat, rye, oats, or barley. A protein of wheat called gluten, and similar proteins in the other grains, causes the problem. As a result of damage to the small intestine, nutrients are not properly absorbed from food.

Cerebrovascular accident. A sudden loss of consciousness resulting when the rupture or blockage of a blood vessel leads to oxygen lack in the brain.

Cheilosis. A painful condition where cracking at the corners of the mouth occurs.

Chronic hepatitis. Mild liver inflammation (swelling and irritation) that may be caused by various viruses and conditions.

Clubbing of the nail beds. An increase in the curvature of the nails so that the surface resembles the back of a teaspoon. Clubbing occurs frequently with cardiopulmonary disease. The change is usually permanent.

Colostomy. The surgical creation of a new opening of the colon on the surface of the body.

Computerized tomography (CT). A series of detailed pictures of areas inside the body taken from different angles; the pictures are created by a computer linked to an X-ray machine.

Congenital heart disease (CHD). A term that includes a variety of structural problems of the heart or its major blood vessels, which are present at birth. Common forms of CHD in Down syndrome are atrioventricular septal defect, atrial septal defect, ventricular septal defect, Tetralology of Fallot, and persistent patent ductus arteriosus. See individual terms for more details about these specific disorders.

Congenital heart failure. Heart failure existing at or before birth.

Congestive heart failure. Prolonged impairment of the ability of the heart to maintain an adequate flow of blood to body tissues and organs.

Co-morbid condition. Condition existing simultaneously with another condition.

Continuous positive airway pressure (CPAP). A term for the device used to treat sleep apnea (stopping of breathing during sleep). This device delivers positive airway pressure at a constant, continuous pressure to help maintain an open airway, allowing a patient to breathe normally through their nose and airway.

Coronary vessels. The vessels that supply the heart muscle with blood rich in oxygen. They are called the coronary arteries because they encircle the heart in the manner of a crown.

Cortical blindness. Loss of sight due to an organic lesion in the visual cortext.

Crypt hyperplasia. The term crypt refers to the mucosal glands of the small intestine. These lie within the connective tissue that lies beneath the epithelium that lines the small intestine. The term hyperplasia refers to a nontumourous increase in the usual number of cells in an organ or tissue. Crypt hyperplasia is characteristic of celiac disease.

Cyanosis. A dark blue to purple discoloration of the skin and mucous membranes that occurs when there is not enough oxygen in the blood (i.e., blood concentration of deoxygenated hemoglobin exceeds approximately 50 grams per litre).

Deep venous thrombosis prophylaxis. Deep vein thrombosis refers to formation of blood clot in a large vein usually in the legs and/or pelvis. Prophylaxis means “the prevention of”. The wearing of surgical knee-high stockings, moving the toes, and taking anticoagulants, can help to prevent blood clot formation.

Dementia. A usually progressive condition marked by the development of multiple cognitive deficits (including memory impairment, aphasia, and inability to plan and initiate complex behavior). Alzheimer disease is the most common form of dementia in the elderly.

Dentition. The kind, number and arrangement of teeth, or growth of teeth .

Dermatitis. Inflammation of the skin.

Diabetes mellitus. A disorder of metabolism characterized by excessively high levels of glucose in the blood. Glucose is a major fuel for the body. The entry of glucose into cells is dependent upon the availability of insulin. In diabetes mellitus, the body does not make enough insulin or has a problem responding to it. Insulin is made specifically by the beta cells in the pancreas. If the beta cells degenerate, type 1 diabetes results. Type 1 diabetes is controlled by the injection of insulin into the body. In type 2 diabetes, the beta cells produce insulin, but there may not be enough of it, and / or cells throughout the body do not respond normally to it. Type 2 diabetes can be controlled by diet, or by oral medicines, but may require insulin injection. By reducing the concentration of glucose in the blood, insulin is thought to prevent or reduce the long-term complications of diabetes, including damage to the blood vessels, eyes, kidneys and nerves.

Dilatation. The widening or expansion of a hollow organ or cavity.

Duodenum. The first part of the small intestine.

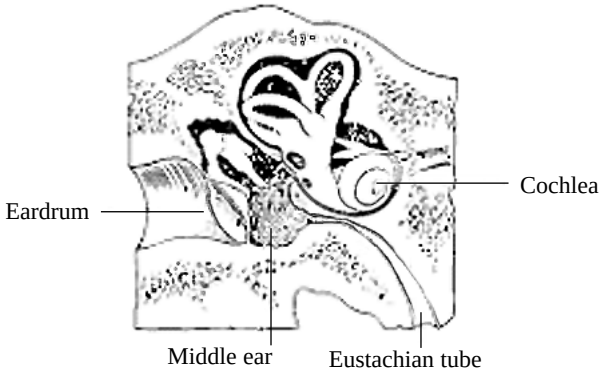
Dobutamine stress echocardiograph. An alternative to the exercise stress test. The drug dobutamine increases the heart rate and blood pressure similar to the effect of exercise.

Dyspnea. Shortness of breath upon exertion.

Dyspepsia. A broad term that includes a variety of digestive problems such as stomach discomfort, gas, bloating, belching, appetite loss, and nausea. Although many serious medical conditions can cause digestive distress, the term "dyspepsia" is most often used when no identifiable medical cause can be detected.

Ear. The ear has three main parts: the outer, middle and inner. The outer ear opens into the ear canal. The eardrum separates the ear canal from the middle ear. Three small bones in the middle ear (the hammer, anvil and stirrup) help transfer sound to the inner ear. The inner ear contains the auditory (hearing) nerve which leads to the brain. Any sound source sends vibrations (sound waves) into the ear. These are channeled through the ear opening, down the ear canal, and strike the eardrum, causing it to vibrate. The vibrations are passed to the small bones of the middle ear, which transmit them to the auditory nerve in the inner ear. Here, the vibrations are changed into nerve impulses that go directly to the brain, which then interprets the impulses as sound (e.g., a voice, a fire alarm, music). Otitis media is an infection of the middle ear behind the ear drum.

Figure 2. Cross-section of the human ear



Medical Illustration Library, 1994 (GA2_6001), by permission.

Echocardiogram (Echocardiograph). A record produced by echocardiography, the use of ultrasound waves, to investigate the action of the heart.

Echocardiography. The ultrasound examination of the heart. A probe which generates harmless sound waves is pressed against the chest. The sound waves bounce off the heart and reconstruct a moving picture of it. The procedure is painless and can help identify abnormalities of the heart muscle and valves as well as identifying fluid around the heart.

Eisenmenger('s) syndrome. A problem of heart function which includes three defects: a defect of the interventricular septum (the barrier between the two ventricles of the heart) in combination with severe pulmonary hypertension (see below), enlargement of the right ventricle, and mild or severe cyanosis. See "Heart" for details of normal heart function.

Electrocardiogram (ECG). A record produced by an electrocardiograph, used in the diagnosis of heart disease. The electrocardiograph is an instrument that displays the electric activity of the heart by means of electrodes attached to the skin.

Endocrine. The endocrine system consists of the glands (and certain tissues) that secrete hormones internally directly into the lymphatic system or the blood stream to control basic body functions such as metabolism, growth and sexual development. The hormones act as “messengers”. They travel to distant tissues and organs to elicit specific functions necessary for the organism. Examples of endocrine glands are the thyroid, parathyroid, adrenals, pancreas and pituitary. See "thyroid" for a diagram of the location of the thyroid and parathyroid glands.

Endocrine disorders. Disorders of the endocrine system. Common examples of endocrine disorders are hypothyroidism, hyperthyroidism, and diabetes.

Endomysium. The delicate connective tissue surrounding the individual muscular fibers within the smallest bundles.

Endoscopy. A procedure for visualizing the interior of a hollow organ or part (as the bladder or esophagus) for diagnostic or therapeutic purposes. This is usually done using an illuminated flexible fiber-optic instrument or a rigid tubular instrument that typically has one or more channels to enable instruments such as forceps or scissors to be passed through.

Enuresis. Inability to control the flow of urine and involuntary urination.

Epicanthic fold. Vertical fold of skin on either side of the nose, sometimes covering the inner canthus (see above). It is present as a normal characteristic in persons of certain races and sometimes occurs as a congenital anomaly in others.

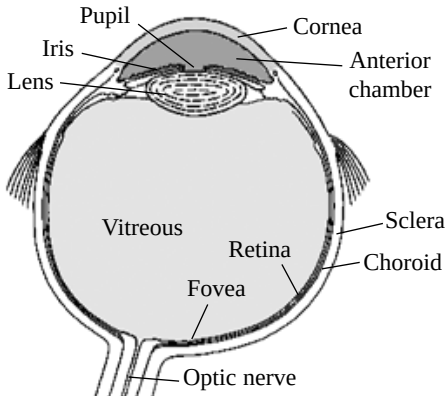
Exocrine. Relating to an organ or gland that secretes substances to its surface via a canal or a duct. Examples of exocrine glands are the salivary glands, the bile producing glands of the liver, the prostate, portion of the pancreas that secretes pancreatic fluid into the duodenum, gastric glands and sweat glands.

Ganglion cells. A group of nerve cell bodies located outside of the central nervous system.

Erythrocytosis. An unexplained chronic disorder characterized by an abnormal increase in red cell number.

Eye. The eye has a number of individual components that are essential for clear vision. The cornea is the transparent outer layer of the front part of the eye; it is the primary focusing element. The outermost layer of the cornea is called the epithelium; this protects the eye. The epithelial cells of the cornea can regenerate quickly. The inner layers of the cornea also are transparent, and allow light to pass. The pupil is the dark opening in the center of the colored iris; this controls the amount of light entering the eye. The iris functions like the iris of a camera; it opens and closes to control the amount of light entering through the pupil. The lens is located just behind the iris; it focuses light rays upon the retina. In young people, the lens is soft and pliable; in older people, it is less pliable, resulting in difficulty in focusing upon objects near to the eye. The retina is the membrane lining the back of the eye; it contains nerve cells which function as photoreceptors. These nerve cells react to the presence and intensity of light by sending an impulse to the brain via the optic nerve. In the brain, the many nerve impulses received from the photoreceptor cells in the retina are formed into an image.

Figure 3. The eye



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Exercise stress test. This is a general screening tool to test the effect of exercise on the heart and give a general sense of how healthy the heart is. It involves measuring the electrical activity of the heart while walking on a treadmill or pedalling on a stationary bicycle. This measures the heart's reaction to the body's increased demand for oxygen. An electrocardiogram (ECG) records the activity of the heart and blood pressure readings are taken. The test continues until a target heart rate is reached. It is stopped if complications such as chest pain or an exaggerated rise in blood pressure develop with activity. Monitoring continues after exercise for 10-15 minutes, or until the heart rate returns to baseline.

Fecal antigen test. A stool test to detect the presence particular proteins or substances (called antigens). One example of such a test is that to detect *Helicobacter pylori* microorganisms.

Gastric cancer. This term refers to several different types of cancer that can occur in the stomach. The most common type is called adenocarcinoma, a form of carcinoma that originates in glandular tissue. To be classified as adenocarcinoma, the cells don't necessarily need to be part of a gland, as long as they have secretory properties. *Helicobacter pylori* infection is a risk factor for gastric cancer.

Gastritis. Inflammation of the stomach, especially of the mucous membrane.

Gastroesophageal reflux disease (GERD). Sometimes called "acid reflux". This is a condition in which the liquid content of the stomach regurgitates (backs up, or refluxes) into the esophagus. The liquid can inflame and damage the lining of the esophagus although this occurs in a minority of patients.

Genetic linkage. This is a term used in genetics and molecular biology. It refers to how close two particular genes or other regions of DNA on a chromosome are to each other.

Glossitis. An abormality of the tongue resulting from inflammation.

Glossoptosis. Abnormal downward or back placement of the tongue.

Glucose. A simple sugar that is the main source of energy in the body. Glucose is the principal sugar the body makes. The body makes glucose from proteins, fats and, in largest part, from carbohydrates. Glucose is carried to each cell through the bloodstream. Cells, however, cannot use glucose without the help of insulin. Glucose is also known as dextrose.

Gluten. A gluey protein substance, especially of wheat flour, that causes dough to be sticky. It is the molecule that triggers celiac disease.

Granulocytes. White blood cells (leukocytes), manufactured in the bone marrow, that contain granules. They destroy and digest bacteria.

Grommets. Ear tube placements to aid with drainage of fluid from the middle ear.

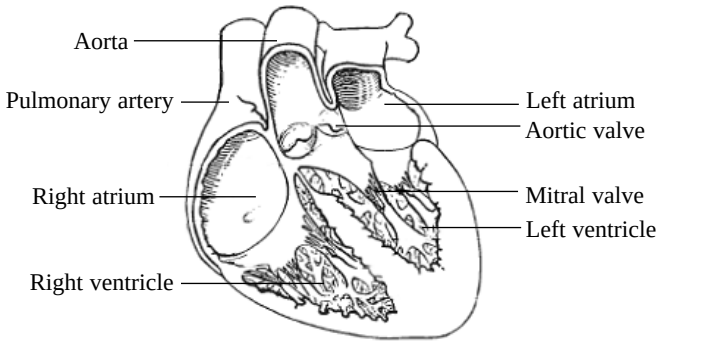
Guaiacum. Substance used in various tests because of the formation of a blue color on oxidation.

Guaiac test. A test for blood in urine or feces using a reagent containing guaiacum that yields a blue color when blood is present. See also *Hemoccult.*

Heart. The muscle that pumps blood received from veins into arteries throughout the body. It is positioned in the chest behind the sternum (breastbone; in front of the trachea, esophagus, and aorta; and above the diaphragm muscle that separates the chest and abdominal cavities. The normal heart is about the size of a closed fist, and weighs about 10.5 ounces. It is cone-shaped, with the point of the cone pointing down to the left. Two-thirds of the heart lies in the left side of the chest with the balance in the right chest. The heart has four chambers - two atria (the upper, low pressure chambers) and two ventricles (the lower, high pressure chambers). With each heartbeat, the atria contract first, pumping blood into the ventricles. Then both ventricles contract, pumping blood out of the heart into the arteries. One-way valves in the heart ensure that the blood flows in the correct direction. The muscular wall called the septum ensures that blood does not leak from one side of the heart to the other. Normally, oxygen-poor blood returns to the right atrium from the body, travels to the right ventricle, and then is pumped via the pulmonary artery into the lungs where it receives oxygen. Oxygen-rich blood returns to the left atrium from the lungs, passes into the left ventricle, and then is pumped out to the body through the aorta (see Figure 4, next page).

Helicobacter pylori. *Helicobacter pylori (H. pylori)* is a bacterial organism responsible for most ulcers and many cases of chronic gastritis. This organism can weaken the protective coating of the stomach and duodenum and allow the damaging digestive juices to irritate the sensitive lining of these body parts. Many people have this organism in their gastrointestinal tract but don't get an ulcer or gastritis. It seems that other factors must also be present for the damage to take place.

Figure 4. The heart



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Hematocrit. A measurement of the proportion of blood that is made up of red blood cells.

Hemoccult. Relating to or being a modified guaiac test in which filter paper impregnated with guaiacum turns blue if occult blood is present.

Hemoptysis. Coughing up of blood or bloody sputum from the lower respiratory tract.

Hepatitis. Inflammation of the liver from any cause. It most commonly is caused by one of three viruses: hepatitis A, B and C. Vaccination can protect against viral hepatitis.

Hepatitis A virus infection. This is spread by the fecal-oral route through close person-to- person contact or by ingesting contaminated food or water. Symptoms can include fever, fatigue, loss of appetite, nausea, abdominal discomfort, jaundice (yellow appearance of the skin and eyes) and dark urine.

Hepatitis B virus infection. This is spread by transmission through blood, semen, vaginal fluids, saliva, breast milk, urine and feces. All individuals who are chronically infected may be able to transmit it. Many people have no noticeable symptoms. Symptoms may develop slowly and include lack of appetite, abdominal discomfort, nausea and vomiting, sometimes joint pain and rash, often progressing to jaundice. Fever may be absent or mild. Incubation period is 45-180 days, rarely as long as 9 months, averaging 60-90 days. Approximately 2-10% of adults and 25-80% of children under the age of 5 will not be able to clear the virus in six months following infection and are considered to

be chronically infected. Long term, chronic hepatitis B can cause liver cell damage, leading to cirrhosis and cancer.

Hepatitis C virus infection. Transmission is primarily through infected blood or blood products. The primary current route of transmission is injection drug use. It is not easily transmitted through sex. In about 10-20% of cases, transmission route has not been identified. 70-75% of people have no noticeable symptoms. Symptoms may develop gradually with loss of appetite, fatigue, abdominal discomfort, nausea and vomiting, sometimes progressing to jaundice (which results in a yellow appearance of skin and eyes). Incubation period ranges from 2 weeks to 6 months, averaging 6-9 weeks. Up to 85% of people with hepatitis C develop chronic infection (which is often asymptomatic). Some develop cirrhosis (scarring of the liver). Chronic hepatitis C infection also substantially increases risk for liver cancer.

HepBeAg (Hepatitis B virus envelope (e) antigen). This is a protein made by hepatitis virus B. The first viral protein to appear in this infection is HepBsAg (see below). Then another antigen named as the hepatitis B envelope (e) antigen (HepBeAg) appears. Traditionally, the presence of HepBeAg in the serum is associated with high rates of viral replication; however, some variants of the hepatitis B virus do not produce the 'e' antigen at all. During the natural course of an infection, antibodies to the 'e' antigen will arise. This is usually associated with a dramatic decline in viral replication.

HepBsAg (Hepatitis B virus surface antigen). This is the first detectable viral protein to appear during infection with hepatitis virus B. Detection of this marker in blood is used most frequently as an indicator of hepatitis B infection. However, early in an infection, this antigen may not be present and it may be undetectable later in the infection as it is being cleared by the host.

HLA type. This term refers to a set of proteins called histocompatibility antigens that are on the surface of the cells in the body. Their main function is to help the immune system defend against invaders such as bacteria, viruses, and parasites. They also are involved in recognizing as foreign the histocompatibility antigens of other people's cells and will fight them, causing rejection of grafts. Histocompatibility antigens in humans are called HLA (for Human Leukocyte Antigens). They are produced by a group of genes in the human major histocompatibility complex (MHC) on human chromosome 6. Certain combinations of HLA antigens predispose to diabetes and to celiac disease.

Hormone. A chemical substance produced in the body that controls and regulates the activity of certain cells or organs. Many hormones are secreted by specialized glands such as the thyroid gland. Hormones are essential for every activity of daily living, including the processes of digestion, metabolism, growth, reproduction, and mood control. Many hormones, such as the neurotransmitters, are active in more than one physical process.

Hydration. The act or process of combining or treating with water; the introduction of additional fluid into the body.

Hypercholesterolemia. Is the presence of abnormally large amount of cholesterol in circulating blood. About 80% of total body cholesterol is made by the liver. The other 20% is from dietary sources. The body produces cholesterol because it is necessary to digest food. Cholesterol is thought to be problem when there is too much "bad cholesterol" or low-density lipoproteins (LDL) present in the body. Excess LDL can trigger the formation of plaque or blockage on the walls of the arteries. Hypercholesterolemia is diagnosed using special blood tests.

Hypersalivation. Excessive salivation.

Hypertension. Abnormally high arterial blood pressure that is usually indicated by an adult systolic blood pressure of 140 millimetres of mercury (mm Hg) or greater or a diastolic blood pressure of 90 mm Hg or greater. Systolic pressure is the blood pressure when the heart is contracting. Specifically it is the maximum arterial pressure during contraction of the left ventricle of the heart. Diastolic pressure refers to the blood pressure when the heart is relaxed. The cause of hypertension is usually not known. It may result in heart disease, stroke, heart failure, kidney disease and blindness. The ultimate consequence of long term high blood pressure could be death.

Hyperviscosity. Slowing or blockage of blood flow due to the increased red cell count.

Hypopharynx. This is the bottom part of the pharynx, and is the part of the throat that connects to the esophagus.

Hypoplasia. Underdeveloped tissue mass.

Hypotension. Abnormally low pressure of the blood; called also *low blood pressure*.

Hypotonic. Lacking in muscle tone.

Hypoxic encephalopathy. Damage to cells in the central nervous system (the brain and spinal cord) from inadequate oxygen.

Ig. Abbreviation for immunoglobulin.

IgA. Abbreviation for immunoglobulin A – a class of immunoglobulins that include antibodies found in external bodily secretions (such as saliva, tears and sweat).

IgM. Abbreviation for immunoglobulin M – a class of immunoglobulins of high molecular weight that include the primary antibodies released into the blood early in the immune response to be replaced later by IgGs of lower molecular weight and that are highly efficient in binding complement.

Immunoglobulin. A protein used by the immune system to identify and neutralize foreign objects like bacteria and viruses. Each antibody recognizes a specific target. Production of antibodies is referred to as the humoral immune system.

Immunologic tolerance. The major mechanism by which tumors grow unchecked by the immune system.

Imperforate anus. Anus lacking the regular opening.

Incentive spirometry. This is a procedure that helps patients to practice and improve their breathing. It encourages the taking of long, slow breaths. It may be ordered after surgery to the abdomen, lungs, neck, or head.

Inflammation. A basic way in which the body reacts to infection, irritation or other injury, the key feature being redness, warmth, swelling and pain. Inflammation is now recognized as a type of nonspecific immune response.

Inflammatory bowel disease. A term encompassing a number of chronic inflammatory disorders leading to damage of the gastrointestinal tract. The most common of these disorders are ulcerative colitis (an inflammation of the large intestine that causes swelling, ulcerations (open sores), and loss of function) and Crohn's disease (an inflammatory bowel disease that causes inflammation or ulceration of the digestive tract; as of 2006, it is suspected that a bacterium which is related to tuberculosis causes Crohn's).

Influenza. A highly infectious disease that affects the respiratory (breathing) tract. It is also known as the flu or grippe. The disease is caused by different types of a virus. When inhaled, the virus attacks

cells in the upper part of the respiratory system and causes symptoms such as fatigue, fever and chills, a hacking cough, and body aches. Influenza can also lead to other, more serious infections.

Insulin. A natural hormone made by the pancreas that controls the level of the sugar glucose in the blood. Insulin permits cells to use glucose for energy. Cells cannot utilize glucose without insulin.

Interproximal bone loss. Interproximal refers to the area between two adjacent teeth. Interproximal bone loss refers to loss of jaw bone in the area between the two teeth.

Intubation. Insertion of a tube into the windpipe through the nose or mouth in order to keep the airway open in patients who are unconscious or who cannot breather for themselves.

Isotonic saline. A 0.89% or 0.90% w/v solution of sodium chloride which is close to the concentration in the blood (isotonic).

Jaundice. Also known as icterus. This is yellowing of the skin, the white of the eyes and mucous membranes caused by increased levels of bilirubin in the human body. Bilirubin results from the breakdown of hemoglobin in worn out red blood cells. Interestingly, it is a potent antioxidant. Jaundice comes from the French word jaune, meaning yellow.

Keratoconus. A degenerative non-inflammatory disorder of the eye in which the cornea thins and changes shape from rounded to more conical. Keratoconus can cause the vision to become distorted quite badly, with multiple images, streaking and sensitivity to light all often reported by the patient. In most cases, corrective lenses enable the affected person to function normally. Occasionally, surgery is necessary.

Lassitude. A condition of weariness, debility, or fatigue.

Lipid profiles. The lipid profile is a group of tests that are often ordered together to determine risk of coronary heart disease. The tests that make up a lipid profile are tests that have been shown to be good indicators of whether someone is likely to have a heart attack or stroke caused by blockage of blood vessels. Tests that are included in a lipid profile include: total cholesterol, HDL-cholesterol (often called good cholesterol), LDL-cholesterol (often called bad cholesterol), and triglycerides. Sometimes the report will include additional calculated values such as HDL/cholesterol ratio or a risk score based on lipid profile results, age, sex, and other risk factors. The lipid profile is used to guide providers in deciding how a person at risk should be treated. The results of the lipid profile are considered along with other known

risk factors of heart disease to develop a plan of treatment and follow-up. See also *Hypercholesterolemia*.

Lymphatic system. The tissues and organs that produce, store, and carry white blood cells that fight infection and other diseases. This system includes the bone marrow, spleen, thymus and lymph glands and a network of thin tubes that carry lymph and white blood cells into all the tissues of the body.

Lymphoma. A usually malignant tumor of lymphoid tissue.

Macroglossia. This term means large tongue. Judging tongue size is subjective; there are no measurements or scale for assessing tongue size. A tongue that protrudes from the mouth, seems too big for the mouth, requires voluntary action to keep it in the mouth and/or results in deformity of the jaws and teeth, is called macroglossia.

Malabsorption. Faulty absorption of nutrient materials from the alimentary canal (passageway from mouth to the anus).

Malignant. Tending to produce death or deterioration.

Meconium. Dark green mucilaginous substance present in the intestine of the full-term baby.

Melena. Vomit or stools stained black by blood pigment. This is usually an indication of bleeding in the upper part of the alimentary canal and especially the esophagus, stomach, and duodenum.

Metabolism. Metabolism is the chemical activity that occurs in cells which releases energy from nutrients or using energy to create other substances, such as proteins. The basal metabolic rate (BMR) is a measurement of energy required to keep the body functioning at rest. Measured in calories, metabolic rates increase with exertion, stress, fear, and illness.

Microsomes. Small, finely granular elements within a cell.

Mitral regurgitation. Leakage of blood back from the lower to the upper chamber of the heart in a severe mitral valve prolapse. See also *Heart*.

Mitral valve prolapse. Billowing of one or both mitral valve leaflets into the left atrium at the end of each active contraction of the heart. The mitral valve controls blood flow on the left side of the heart. The valve opens and closes with each heartbeat. It works like a one-way gate, letting blood flow from the upper heart chamber to the lower chamber.

When the mitral valve is prolapsed, it bulges like a parachute; if the prolapse is severe, then blood may leak back from the lower to the upper chamber. See also *Heart.*

Morbidity. The incidence of disease or the rate of sickness in a specified community or group.

Mortality. The number of deaths in a given time or place; or, the proportion of deaths to population. The death rate is also called mortality rate.

Magnetic resonance imaging (MRI). A form of medical imaging that uses a combination of radio frequency energy and a powerful magnetic field.

Mucous membrane. A thin layer of tissue or membrane that lines the interior surface of body openings and secretes mucous.

Myelofibrosis. Increase in the amount of fibrous tissue in the bone marrow.

Myoclonus. Jerking, involuntary movements of the arms and legs; may occur normally during sleep.

Murmur. An atypical sound (e.g., of the heart) typically indicating a functional or structural abnormality.

Neurofibrillary tangles. These are insoluble twisted fibres known as known as paired helical filaments; they are found within the nerve cells (neurons) in certain neurodegenerative diseases such as Alzheimer's, and are thought to disrupt cell function. They primarily consist of a protein called tau, which forms part of a structure called a microtubule. The microtubules help transport nutrients and other important substances from one part of the neuron to another. In Alzheimer disease, the tau is abnormal (it carries too many phosphate groups) and the microtubule structures collapse. Because neurofibrillary tangles will bind the metal ions iron and aluminum, some researchers believe that neurofibrillary tangles also play a role in defending the brain against metal ion toxicity (Brown & Percy, 2003, pp. 793-807, by permission).

Neutrophils. One of the two most common types of white blood cells that often predominates in the earliest stages of an infection; these tend to be short-lived compared to other types of white blood cells.

Obesity. Condition that is characterized by excessive accumulation and storage of fat in the body and that in an adult is typically indicated by a body mass index of 30 or greater.

Occlusal. Pertaining to the contacting or biting surfaces of opposing teeth.

Optometrist. An eye care professional, but not a medical doctor.

Opthalmologist. Ophthalmology is the branch of medicine which deals with the diseases of the eye and their treatment. An opthalmologist is a medical doctor specially trained in this field.

Osteoporosis. This term means porous, fragile bones. It is a disease in which there is exaggerated loss of quantity and quality of bone, causing an increase in the risk of fractures.

Otoscopy. Examination of the ear with an otoscope (this consists essentially of a magnifying glass and a light).

Palliative care. The term palliative care refers to an approach that improves the quality of life of patients and their families facing the problem associated with life-threatening illness, through the prevention and relief of suffering by means of early identification and impeccable assessment and treatment of pain and other problems, physical, psychosocial and spiritual.

Palmoplantar hyperkeratosis. Skin disorder primarily affecting the palms of the hands and soles of the feet.

Palpebral fissures. The gap between the upper and lower eyelids. Palpebral means pertaining to the eyelids. The term fissure refers to the space between the eyelids when the eye is open.

Patent ductus arteriosus (PDA). A connection between the aorta and the pulmonary artery which allows oxygen-rich blood that should go to the body to recirculate through the lungs.

Pancreas. An organ that serves two functions:

- exocrine – it produces pancreatic juice containing digestive enzymes.
- endocrine – it produces several important hormones, including insulin.

Peptic ulcer disease. Ulcers are erosions (wearing away or corrosion) in the lining of the stomach or duodenum. An ulcer in the stomach is called a gastric ulcer. An ulcer in the duodenum is called a duodenal ulcer. Together, ulcers of the stomach and duodenum are referred to as peptic ulcers.

Persistent patent ductus arteriosus. Patent ductus arteriosus (PDA) is a congenital heart disease. A PDA is a persistent connection between the aorta and the pulmonary artery. This connection is called the ductus

arteriosus and is normally present before birth. In most babies, the vessel closes within a few hours to days after birth. In some children, this vessel fails to close.

Phlebotomy. Phlebotomy is the act of drawing or removing blood from the circulatory system through a cut (incision) or puncture in order to obtain a sample for analysis and diagnosis. Phlebotomy is also done as part of the patient's treatment for certain blood disorders.

Physiotherapy. The treatment of disease by physical and mechanical means (as massage, regulated exercise, water, light, heat, and electricity).

Pneumococcus. A bacterium of the genus Streptococcus (S. pneumoniae) that causes an acute pneumonia involving one or more lobes of the lung.

Pneumonia. A disease of the lungs that is characterized especially by inflammation and consolidation of lung tissue followed by resolution, is accompanied by fever, chills, cough, and difficulty in breathing, and is caused chiefly by infection.

Polycytemia. A condition marked by an abnormal increase in the number of circulating red blood cells.

Polymorphic variant. A naturally occurring or induced variation in the sequence of genetic information (i.e., nucleotide bases) on a segment of DNA.

Polysomnography. The technique or process of using a polygraph to make a continuous record during sleep of multiple physiological variables (as breathing, heart rate, and muscle activity).

Presbyacusis. Losing one's hearing as one grows older – the most common type of deafness.

Prevalence. The percentage of a population that is affected with a particular disease at a given time.

Primary sclerosing cholangitis. In primary sclerosing cholangitis (PSC), the bile ducts inside and outside the liver become inflamed and scarred. As the scarring increases, the ducts become blocked. The ducts are important because they carry bile out of the liver. Bile is a liquid that helps break down fat in food. If the ducts are blocked, bile builds up in the liver and damages liver cells. Eventually, PSC can cause liver failure.

Pruritis. Localized or generalized itching due to irritation of sensory nerve endings.

Pulmonary. Relating to, functioning like, associated with, or carried on by the lungs.

Pulmonary artery. An arterial trunk or either of its two main branches that carry oxygen-deficient blood to the lungs.

Pulmonary hypertension (PAH). Is continuous high blood pressure in the pulmonary artery. The average blood pressure in a normal pulmonary artery is about 14 millimetres of mercury (mmHg) when the person is resting. In PAH, the average is usually greater than 25 mmHg. PAH begins when tiny arteries in the lungs become narrow or blocked. This causes increased resistance to the flow of blood in the lungs, which in turn raises pressure within the pulmonary arteries. As the pressure builds, the heart's lower right chamber (right ventricle) must work harder to pump blood through the lungs, eventually causing the heart muscle to weaken and sometimes to fail completely. Although pulmonary hypertension isn't curable, treatments are available that can help lessen symptoms and improve quality of life for many people with pulmonary hypertension.

Pulmonary pressure. This refers to the blood pressure in the pulmonary arteries.

Pulmonary vasculature. This term refers to the veins, arteries and capillaries that comprise the circulatory system of the lungs. Because pulmonary circulation has lower pressure than systemic circulation, pulmonary arteries and veins tend to have more delicate walls than those seen elsewhere in the body.

Reagent. A substance used to test or measure a component because of its chemical or biological activity.

Refractive error. This means that shape of the eye is such that it does not bend light correctly, and the image is blurred. Light has to be refracted or bent by the cornea and the lens to the retina in order for us to see. The common refractive disorders are myopia (distant objects are blurry), hyperopia (close objects are blurry), presbyopia (aging of the lens in the eye), and astigmatism (blurred vision at all distances). See also *Eye*.

Refractory celiac disease. Celiac disease that does not respond to a gluten-free diet; this occurs rarely.

Regurgitation. A backward flow, as of blood, through an incompetent valve of the heart.

Respiratory centre. The respiratory centre is the region of the brain medulla that controls breathing. It receives inputs from various receptors around the body and sends output through two nerves to the muscles around the lung. The medulla and its nerves are part of the autonomic (involuntary) nervous system.

Respiratory effort. One category of the APGAR score:

- If there are no respirations, the infant scores 0 for respiratory effort.
- If the respirations are slow or irregular, the infant scores 1 for respiratory effort.
- If there is good crying, the infant scores 2 for respiratory effort.

Secondary reduction of chest wall expansion. During quiet breathing, the rib cage normally expands symmetrically as the lungs fill with air. With aging, there can be a decline in elasticity of the rib cage, and a loss of muscle mass that results in weakening of the muscles of respiration.

Secundum atrial septal defect. A particular type of atrial septal defect in which there is an abnormally large opening in the atrial septum at particular sites: the foramen ovale and the ostium secundum.

Seizures. Convulsions, body spasms or shaking resulting from abnormal electrical discharges in the brain.

Sensitivity. In tests that provide a yes or no answer, such as a blood test to determine if a person has or has not a particular disease, the test sensitivity is defined as the proportion of those cases having a positive test result of all positive cases (e.g., people with the disease) tested. It is defined as the number of true positives divided by the number of true positives plus the number of false negatives. A sensitivity of 100% means that all sick people are recognized as such.

$$\text{sensitivity} = \frac{\text{number of true positives}}{\text{number of true positives} + \text{number of false negatives}}$$

Serological testing. Serology is a medical blood test to detect the presence of antibodies against a microorganism, such as antibodies produced against the envelope antigen of hepatitis B virus. A serology test may be performed when an infection is suspected. There are several serology techniques that can be used depending on the suspected antibodies. Serology techniques include agglutination, precipitation, complement-fixation and fluorescent antibodies.

Sleep apnea. Brief periods of recurrent cessation of breathing during sleep that is caused especially by obstruction of the airway or a disturbance in the brain's respiratory center and is associated with excessive daytime sleepiness.

Spasms. Sudden, involuntary muscle contractions.

Specificity. In testing that yields yes or no answers (as in a diagnostic test for a certain disease), specificity is the proportion of true negatives of all the negative samples tested, that is number of true negatives divided by the number of true negatives plus number of false positives. For a test to determine who has a certain disease, a specificity of 100% means that all healthy people are labelled as healthy. Specificity alone does not tell us all about the test. We also need to know the sensitivity of the test.

$$\text{specificity} = \frac{\text{number of true negatives}}{\text{number of true negatives} + \text{number of false positives}}$$

Sputum. The matter discharged from the air passages in diseases of the lungs, bronchi, or upper respiratory tract that contains mucus and often pus, blood, fibrin, or bacterial products.

Steathorrea. An excess of fat in the stools.

Stomatitis. Inflammation of the mouth.

Stroke. A stroke is the sudden death of cells in a limited part of the brain caused by a reduced flow of blood to the brain. Reduction of blood flow can be caused by blood clots that form in the brain, blood clots that travel from other parts of the body to the brain, or by the breakage of blood vessels in the brain.

Swallowing disorder. Also called dysphagia, or impaired swallowing. Swallowing is a complex act that involves the mouth, throat area, and esophagus (tube that transports food to the stomach). Many nerves and muscles affect the correct function of these parts. Part of the act of swallowing is under conscious (voluntary) control. However, much of swallowing is involuntary. There are many different causes of swallowing difficulty. These can be distinguished with a thorough medical history, physical exam, and testing. They include: obstruction to the passage of food or liquid; nerve and muscle problems; and, problems related to the esophagus.

Syncope. Loss of consciousness resulting from insufficient blood flow to the brain (fainting).

Syncopal episodes. Episodes of fainting.

Syndrome. A group of signs and symptoms that occur together and characterize a particular abnormality.

Syntactic. Pertaining to syntax (see below).

Syntax. The grammatical arrangement of words, showing their connection and relation.

Systemic circulation. This supplies nourishment to all tissues of the body with the exception of the heart and lungs, because they have their own circulatory systems.

Systemic vascular resistance. Resistance offered by the peripheral circulation.

Tetralogy of Fallot. A congenital abnormality of the heart characterized by four key features. A ventricular septal defect (a hole between the ventricles) and many levels of obstruction from the right ventricle to the lungs (pulmonary stenosis) are the most important. Also, the aorta (major artery from the heart to the body) lies directly over the ventricular septal defect, and the right ventricle develops thickened muscle.

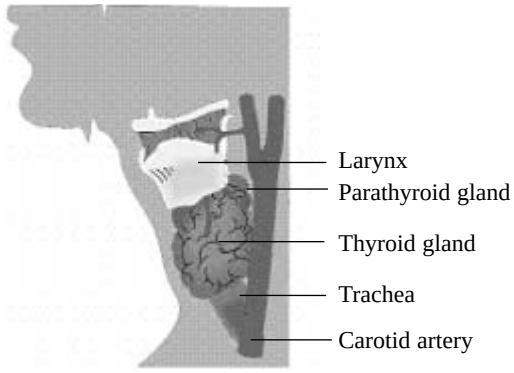
Thalamus. A part of the brain. The two thalami are located in the centre of the brain, one beneath each cerebral hemisphere and next to the third ventricle. Functionally the thalami can be thought of as relay stations for nerve impulses carrying sensory information into the brain; the thalami receive these sensory inputs as well as inputs from other parts of the brain and determine which of these signals to forward to the cerebral cortex.

Thallium persantin test. An alternative to the exercise stress test for patients who cannot exercise on a treadmill. The drug persantin (dipyridamole) helps to expand the coronary arteries increasing the blood flow to the heart. This effect is similar to what happens during vigorous exercise.

Thyroid. A gland that makes and stores hormones that help regulate the heart rate, blood pressure, body temperature, and the rate at which food is converted into energy. Thyroid hormones are essential for the function of every cell in the body. They help regulate growth and the rate of chemical reactions (metabolism) in the body. Thyroid hormones also help children grow and develop. The thyroid gland is located in the lower part of the neck, below the Adam's apple, wrapped around the trachea (windpipe) (see Figure 5). Thyroid hormone production (T3 and T4) is regulated in the following way. The thalamus produces thyroid

releasing hormone (TRH). which acts on the pituitary to produce thyroid stimulating hormone (TSH), also called thyrotropin. TSH stimulates stimulates the thyroid to produce T3 and T4. When T3 and T4 levels rise sufficiently, the release of TRH and TSH is suppressed.

Figure 5. Cross-section of the neck



Corel Gallery 2, 1995, by permission

T3. Triiodothyronine, one of the active circulating hormones of the thyroid.

T4. Thyroxine, the main active circulating hormone of the thyroid.

TRH. Thyroid releasing hormone. This hormone is produced by the thalamus. It stimulates the pituitary to produce TSH.

TSH. Thyroid stimulating hormone or thyrotropin. This hormone is secreted by the pituitary. It stimulates the thyroid to synthesize T3 and T4. When T3 and T4 levels rise sufficiently, the release of TRH and TSH is suppressed.

Tonic clonic seizures. This type of seizure is characterized by sudden loss of consciousness and jerking of the body musculature that may last for up to a few minutes. After a grand mal seizure (post-ictal period), a patient may exhibit various behaviors, ranging from regaining consciousness, confusion, disorientation, absence of memory of the immediate events, to further seizures.

Transglutaminase. A clotting factor that is a variant of factor XIII and that promotes the formation of cross-links between strands of fibrin. Antibodies to transglutaminase are produced in celiac disease.

Transcription factor. Any of various proteins that bind to DNA and play a role in the regulation of gene expression by promoting transcription (production of RNA using DNA as a template).

Urea breath test. This is used to detect the presence of H. pylori in the stomach. A small amount of food containing radioactive carbon is administered. If there are a lot of bacteria in the stomach, they will convert this into radioactive carbon dioxide that will be breathed out. The radioactive carbon dioxide is collected into a bag and the amount of radioactivity measured. The test is used to determine if a patient is infected and should be put on antibiotics, or if they have been treated with antibiotics and essentially cured of the infection.

Valvular disease. Valvular heart disease refers to several disorders and diseases of the heart valves, which are the tissue flaps that regulate the flow of blood through the chambers of the heart.

Ventricle. A chamber of the heart which receives blood from a corresponding atrium and from which blood is forced into the arteries.

Ventricular septal defect. An opening in the ventricular septum of the heart. This allows blood to flow between the left and right sides of the heart.

Villus (plural, villi). A small slender fingerlike process with a blood supply that sticks out from the surface of some types of cells such as those in the small intestine.

Vitamins. Any of various organic substances that are essential in minute quantities to the nutrition of most animals and some plants, act especially as coenzymes and precursors of coenzymes in the regulation of metabolic processes but do not provide energy or serve as building units, and are present in natural foodstuffs or are sometimes produced within the body.

A. Any of several fat-soluble vitamins or a mixture of two or more of them whose lack in the animal body causes keratinization of epithelial tissues (as in the eye with resulting night blindness and xerophthalmia).

D. Any or all of several fat-soluble vitamins chemically related to steroids, essential for normal bone and tooth structure, and found especially in fish-liver oils, egg yolk, and milk or produced by activation (as by ultraviolet irradiation) of sterols.

E. Any of several fat-soluble vitamins that are chemically tocopherols or tocotrienols, are essential in the nutrition of various

vertebrates in which their absence is associated with infertility, degenerative changes in muscle, or vascular abnormalities, are found especially in wheat germ, vegetable oils, egg yolk, and green leafy vegetables or are made synthetically, and are used chiefly in animal feeds and as antioxidants.

K. Either of two naturally occurring fat-soluble vitamins that are essential for the clotting of blood because of their role in the production of prothrombin in the liver and that are used in preventing and treating hypoprothrombinemia and hemorrhage.

Folate. Crystalline vitamin of the B complex that is required for normal production of red blood cells, that is used especially in the treatment of nutritional anemias, and that occurs especially in green leafy vegetables, liver, kidneys, dried beans, and mushrooms – called also folacin, folate, Lactobacillus casei factor, pteroylglutamic acid, vitamin Bc, vitamin M.

B12. A complex cobalt-containing compound that occurs especially in liver, is essential to normal blood formation, neural function, and growth, and is used especially in treating pernicious and related anemias and in animal feed as a growth factor – called also cyanocobalamin.

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Yale Medical Group. *Diabetes & Other Endocrine & Metabolic Disorders.*
http://ymghealthinfo.org/content.asp?page=P01964

Appendix II

More Resources Relevant to Health Issues in Adults With Down Syndrome

Brain Bank

Canadian Brain Tissue Bank

The banking of brain tissue from people with or without Down syndrome is a crucial resource for the study of Down syndrome and one of the most important legacies. The Canadian Brain Tissue Bank has a formal donor plan to retrieve tissue that is scientifically important and to make it available to researchers for peer-reviewed projects.
http://www.utoronto.ca/neuropathology/cbb.html

Books, Journal Articles, Pamphlets and CDs

In addition to the readings in this section, see also Internet Resources for additional information.

Ageing and its consequences for people with Down's syndrome: A guide for parents and carers. (2004). Teddington, UK: Down's Syndrome Association, UK.

Aging with a developmental disability. (2004). (English and French). Ottawa: National Advisory Council on Aging.

Alzheimer's disease in individuals with Down syndrome. (2005). Norberto Alvarez. Boston, MA: Emedicine.com.

The three pamphlets above are available online through the *Alzheimer Society of Toronto*. Many other books and pamphlets also are available as references or loans. (See *Alzheimer Society of Toronto Internet Resource* below.)

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http://www.uic.edu/orgs/rrtcamr/dbiblio.htm

Brown, I., & Percy, M. (Eds.). (2003). *Developmental disabilities in Ontario*. Toronto: Ontario Association on Developmental Disabilities.

This book contains 45 chapters that provide a comprehensive guide to the intellectual and developmental disabilities, extensive reference lists and Ontario-specific resources. In particular, this book contains chapters on health issues in Down syndrome, Alzheimer disease, Alzheimer disease in Down syndrome, and physical health issues of relevance to all persons with intellectual and developmental disabilities.
http://www.oadd.org

Child and adult series of the Down Syndrome Educational Trust.

DSii dult and social issues series. Roy I. Brown, Editor. Portsmouth, UK: Down Syndrome Educational Trust.

DSii education and development series. Sue Buckley, Editor. Portsmouth, UK: Down Syndrome Educational Trust.

http://www.downsed.org/publishing/dsii/

Davidson, P., Prasher V.P., & Janicki, M. (2003). *Mental health issues in intellectual disabilities.* Oxford: Blackwell Publishing.

One of a series of publications designed to address the issues of health, adult development and aging among persons with intellectual disabilities by IASSID (International Association for the Scientific Study of Intellectual Disabilities).
http://www.blackwellpublishing.com/seriesbyseries.asp?ref=SSID

Developmental disabilities and Alzheimer's disease: What you should know. Silver Spring, MD: The Arc of the United States.

http://www.thearc.org/misc/alzbk.html

Down syndrome, issues and interventions, A multi-media CD-ROM for Windows and Mac. (2000). Vancouver, Canada: Down Syndrome Research Foundation & Resource Centre.

This CD-ROM provides an introductory overview of a wide array of issues relevant to people with Down syndrome and several interventions currently being used. Adopting a life-span approach, the information spans the needs of people with Down syndrome from infancy to adulthood.
http://www.videoactive.com

Hawkins, B., & Eklund, S. (1994). *Aging related change in adults with mental retardation.* Research brief. Silver Spring, MD: The Arc of the United States.

http://www.uic.edu/orgs/rrtcamr/aboutus.htm

Janicki, M.P., & Dalton, A.J. (2000). Prevalence of dementia and impact on intellectual disability services. *Mental Retardation, 38,* 277-289.

http://aamr.allenpress.com/aamronline/?request=get-abstract&doi=10.1352%2F0047-6765(2000)038%3C0276:PODAIO%3E2.0.CO%3B2

Janicki, M.P., & Dalton, A.J. (Eds.). (1999). Aging, dementia and intellectual disabilities: A Handbook. Philadelphia: Brunner/Mazel.

This book contains 21 well-referenced chapters that provide information about dementia and aging as they affect people with developmental disabilities. The book also contains a Dementia Assistive Resource Directory with Internet addresses and Appendices that summarize the AAMR-IASSID practice guidelines for diagnosis and care management of adults with intellectual disabilitiy and dementia, guidelines for coping with Alzheimer disease in persons with Down syndrome, and instruments and tests that have been used in the diagnosis of dementia in people with intellectual disability.

http://www.wrongdiagnosis.com/amazon/books/down-syndrome/dementia-and-aging-adults-with-intellectual-disabilities-a--0876309163.html

Janicki, M.P., Heller, T., Seltzer, G., & Hogg, J. (1996). Practice guidelines for the clinical assessment and care management of Alzheimer's disease and other dementias among adults with intellectual disability. *Journal of Intellectual Disability Research, 40*, 374-382.

Also available from the *American Association on Mental Retardation* in print form or on the web:

http://cc.msnscache.com/cache.aspx?q=2965573681680&lang=en-CA&mkt=en-CA&FORM=CVRE4

Jobling, A., & Virji-Babul, N. (2004). Down syndrome: Play, move and grow. Vancouver, Canada: Down Syndrome Research Foundation.

This book, aimed at parents, educators, and physical, occupational, and play therapists, offers a comprehensive overview of motor development in children and adults with Down syndrome. Making the link between motor development and health, the book presents both current research literature and suggestions for developing and promoting motor development through play, recreation, and exercise. A reference list and helpful resources are also provided.

http://www.dsrf.org/

Medlen, J.E. (2002). *The Down syndrome nutrition handbook: A guide to promoting healthy lifestyles.* Bethesda, MD:Woodbine House.

This comprehensive guide provides information relevant to children

and adults with Down syndrome about nutrition and healthy lifestyles. Topics covered include breast and bottle feeding, basic nutrition, diabetes, celiac disease, weight management, fitness, and others. http://www.parentbooks.ca/

Prasher, V.P. (Ed.) (2005). *Alzheimer's disease and dementia in Down syndrome and intellectual disabilities.* Oxford: Radcliffe Publishing.

This book helps carers and professionals who are living or working with adults with Down syndrome and intellectual disability to increase their understanding of Alzheimer's disease and other forms of dementia which can disproportionately affect this patient group. It relates research to clinical practice and shows how early diagnosis, appropriate treatment and compassionate care can be used effectively to maintain dignity and quality of life.
http://www.radcliffe-oxford.com/books/bookdetail.asp?ISBN=1+85775+608+8

Prasher, V.P. (Ed.) (In press). *Down syndrome and Alzheimer's disease: Biological correlates.* Oxford: Radcliffe Publishing. Publication date: July 2006

To date, interest in this area has been focused on the clinical and diagnostic aspects of dementia in Alzheimer's disease [DAD] in the intellectually disabled population. Until recently the underlying biological abnormalities, possibly giving rise to the clinical psychopathology of DAD in persons with Down syndrome, have been neglected. This book is the first book in the field of intellectual disability which has been published to re-address this concern.
http://www.radcliffe-oxford.com/books/bookdetail.asp?ISBN=1+85775+637+1

Prasher, V.P., & Janicki, M.P. (Eds.) (2002). *Physical health in adults with intellectual disabilities. Oxford: Blackwell Publishing.*

One of a series of publications designed to address the issues of health, adult development and aging among persons with intellectual disabilities by IASSID (International Association for the Scientific Study of Intellectual Disabilities).
http://www.blackwellpublishing.com/seriesbyseries.asp?ref=SSID

Prasher V.P., & Smith, B. (2002). *Better healthcare for people with Down syndrome.* Kidderminster, UK: BILD Publications.

This book provides practical information for staff, families and other carers on the important health issues for adults with Down syndrome.
http://www.bild.org.uk/publications/health.htm

Pueschel, S.M. (Ed.). (In press). *Adults with Down syndrome.* Baltimore: Paul H. Brookes Publishing Co. Publication date: July, 2006.

This book deals with the social, clinical, legal, and personal issues people with Down syndrome will navigate in adulthood.
http://www.pbrookes.com/store/books/pueschel-8116/index.htm

Sano, M., Aisen, P.S., Dalton A.J., Andrews, H. F., & Tsai, W.Y. (2005) Assessment of Aging Individuals with Down Syndrome in Clinical Trials: Results of Baseline Measures. *Journal of Policy and Practice in Intellectual Disabilities, 2*, 126-138.

This reference describes the instruments used to measure neurocognitive and neurobehavioural function in the Clinical Trial of Vitamin E in Down Syndrome (see below).

Todd, K, Turk, V., & Christmas, M. (2003). *Down syndrome and dementia resource pack.* Kidderminster, UK: BILD Publications.

A resource for family carers, staff and other professionals supporting people with Down syndrome, to help them with practical day to day issues. See: http://www.bild.org.uk/publications/health.htm

Victoroff, J. (2002). *Saving your brain.* New York:Bantam Doubleday Dell.

This book contains much sensible advice for maximizing mental fitness.
Available from Amazon.com: see
http://www.amazon.com/gp/product/0553109448/ref=br_lf_b_/102-3677249-7156121?n=10&s=books&v=glance

Noonan-Walsh, P., & Heller, T. (Eds.). (2003). *Health of women with intellectual disabilities.* Oxford: Blackwell Publishing.

One of a series of publications designed to address the issues of health, adult development and aging among persons with intellectual disabilities by IASSID (International Association for the Scientific Study of Intellectual Disabilities).
http://www.blackwellpublishing.com/seriesbyseries.asp?ref=SSID

Your good health. Kidderminster, UK: BILD Publications.

12 illustrated booklets to inform people with a learning disability about health issues and explain how to get help for a variety of health issues (Getting Older - Feeling Good, Pregnancy and Child Birth, Epilepsy, Exercise, Breathe Easy, Eating and Drinking, Alcohol and Smoking, Looking After Your Teeth, Seeing and Hearing, Sex, If you are ill, Using Medicine Safely, Coping With Stress).
http://www.bild.org.uk/publications/your_very_good_health_details.htm

Clinical Trials in Aging Persons with Alzheimer Disease
Clinical Trial: Vitamin E in Aging Persons with Down Syndrome.

This international study is currently recruiting patients. The goal of this study is to determine the safety and efficacy of the administration of vitamin E, which has been shown to delay the progression of Alzheimer disease, in slowing the rate of cognitive/functional decline in older persons with Down syndrome.
http://www.clinical trials.gov/ct/gui/show/NCT00056329.
Phone (416) 925-5141, ext. 353, for information about the study in Ontario. Check the website for information about the study in British Columbia, Saskatchewan and other international sites. See Sano et al., 2005, for a description of the instruments being used to measure neurocognitive and neurobehavioural function in the study.

Community Supports

In addition to the resources listed below, see various Down Syndrome Associations and Societies and Alzheimer Disease Associations and Societies in Internet Resources.

Networks of Specialized Care in Ontario. Reshaping Community-Based Specialized Services In Ontario.

This site explains the plan of the Ontario government to better coordinate access to specialized services, improve the way services are delivered and promote professional development through increased sharing of research and training. The eight sites that will serve as "lead agencies" are listed below.
http://www.mcss.gov.on.ca/CFCS/en/newsRoom/backgrounders/060302.htm

Southern Ontario
Bethesda
http://www.bethesdaservices.com/
Regional Supports Associates
http://www.regionalsupport.on.ca/about.html

Central Ontario
Surrey Place Centre
http://www.surreyplace.on.ca/whoweare_overview.php
Community Living Huronia (Pineview Site)
http://www.clhmidland.on.ca/Home.htm
Guelph Community Mental Health Clinic
http://www.freespace.net/~cmhc/

Eastern Ontario
Ongwanada (Kingston)
http://www.ongwanada.com/
Prescott-Russell Services for Children and Adults
http://www.seapr.ca/

Northern Ontario
Algonquin Child and Family Services
http://www.acfs.on.ca/home.php

Clinics

Memory Clinic, Toronto Western Hospital, Toronto
http://www.uhn.ca/programs/clinics/memory.asp
Memory Clinic, Baycrest Centre for Geriatric Care, Toronto
http://www.baycrest.org/Brain_Health_Centre/Memory_Clinic/

Dentistry for Persons with Disabilities, Mount Sinai Hospital, Toronto
http://www.mtsinai.on.ca/Dentistry/default.htm

Rehabilitation Institute of Toronto (Toronto Rehab)
http://www.torontorehab.on.ca/about/index.htm

Other

Safely Home Registry (Wandering Person's Registry)
http://www.alzheimer.ca/english/safelyhome/intro.htm

Canadian Association of Occupational Therapists (CAOT)
http://www.caot.ca/default.asp?pageid=1
Ontario Home Care Association
http://www.homecareontario.ca/public/index.cfm

Ontario Ministry of Health and Long-Term Care
http://www.health.gov.on.ca/index.html

Community Care Access Centres (CCAC)
http://www.health.gov.on.ca/english/public/program/ltc/6_ccac.html

Ontario Ministry of Community and Social Services
http://www.cfcs.gov.on.ca/CFCS/en/default.htm

Roeher Institute

A leading policy-research and development organization. Its mission is to generate knowledge, information and skills to secure the inclusion, citizenship, human rights and equality of people with intellectual and other disabilities.
http://www.cacl.ca/English/aboutus/roeher.html

Internet Resources

Adult Down Syndrome Center of Advocate Lutheran General Hospital

Newsletters prepared by the Center and papers written by staff can be downloaded from this site.
http://www.advocatehealth.com/luth/services/other/adsc/publications.html

Aging with Developmental Disabilities: Women's Health Issues

By Allison A. Brown, Rehabilitation Research and Training Center on Aging with Mental Retardation, University of Illinois at Chicago, Chicago, Illinois; and, Leone Murphy, R.N., The Arc of Monmouth County, Tinton Falls, New Jersey
http://www.thearc.org/faqs/whealth.html

American Association for Mental Retardation (AAMR)

http://www.aamr.org

Best Buddies

http://www.bestbuddies.org

Fact Sheet: Aging.Older Adults and Their Aging Caregivers

AAMR
http://www.aamr.org/Policies/faq_aging.shtml

Canadian Down Syndrome Society

This organization links parents and professionals through advocacy, education, and providing information.
http://www.cdss.ca/en/main.htm#
A site for adults with Down syndrome, including self-advocacy, health, games, and Internet, is under development and will soon be posted.
http://www.reubenhall.com/cdssWebsite/siteForAdults.html

Down's Heart Group

Offers support and information relating to heart conditions associated

with Down syndrome.
http://www.dhg.org.uk/informationpack.htm

Down Syndrome Association of Ontario
This organization is formed of Down Syndrome Associations from across the province. As of 2006, it is focusing on individualized funding and appropriate housing for adults with Down syndrome
http://www.dsao.ca/

Down Syndrome Association of Toronto
This is a non-profit organization providing support and information to parents of children with Down syndrome, students and teachers. The objective of this organization is to make information accessible to as many people as possible around the world.
http://dsat.ca/

Down Syndrome: Health Issues - Medical Essays and Information
Essays on a variety of health issues by Dr. Len Leshin; updated January 19, 2006.
http://www.ds-health.com/

Down Syndrome Information Network
Offers a range of information resources and online services to the international Down syndrome community. It aims to provide information and services for families, carers, professionals and researchers worldwide. It has an extensive online library.
http://www.down-syndrome.info/topics/recommended/families/

Down Syndrome Educational Trust
This site has an extensive list of materials for parents, professionals and educators covering a fairly broad array of issues.
http://wwww.downsed.org/

Down Syndrome Research Foundation (Canada)
This organization was formed in 1995 in response to the need, expressed by parents and professionals, for detailed and research-based information for themselves and the community at large. Though based in British Columbia, it serves people across Canada.
http://www.dsrf.org/
Check the following site for articles, websites and other resources on many subtopics related to health.
http://dsrf.org/index.cfm?fuseaction=inform.medical

Down's Syndrome Scotland

This site reviews many different topics relevant to Down syndrome, including health, aging and dementia and down syndrome.
http://www.dsscotland.org.uk/

Down Syndrome Title Page

A listing of organizations worldwide, support groups, including toy catalogs for children of special needs.
http://www.nas.com/downsyn/

DownSyndrome.com

Directory of Down syndrome Internet Sites (U.S. and international). Go to this site to find the Down Syndrome Association nearest you.
http://www.downsyndrome.com/

DS WWW home page

http://www.nas.com/downsyn/

Information About Mental Retardation and Related Topics

http://www.thearc.org/info-mr.html

Learning About Intellectual Disabilities and Health (UK)

A web-based learning resource for medical and health care students and practitioners; contributions by experts from around the world are already on this website.
http://www.intellectualdisability.info

Mile High Down Syndrome Association

On this site is a large list of books on many different topics relevant to Down syndrome.
http://www.mhdsa.org/LibraryAlpha.htm

National Down Syndrome Congress

http://www.ndsccenter.org

National Down Syndrome Society (U.S.) - Information and Resources

The mission of this organization is to benefit people with Down syndrome and their families through national leadership in education, research and advocacy. Information on many different health topics are available on this site.
http://www.ndss.org/content.cfm?fuseaction=InfoRes.Hlth

National Institutes of Health. National Institute of Child Health and Human Development

Health Information and Media Publications. Facts about Down syndrome.
http://www.nichd.nih.gov/publications/pubs/downsyndrome/down.htm

Recommended Down Syndrome Sites on the Internet

This list has been compiled from among thousands by Dr. Len Leshin; updated February 8, 2006.
http://www.ds-health.com/ds_sites.htm

Riverbend Down Syndrome Parent Support Group

This site has information on different topics relevant to Down syndrome including life planning and medical health.
http://www.altonweb.com/cs/downsyndrome/index.html

Rehabilitation Research and Training Center on Aging With Developmental Disabilities (RRTC)

This web site is designed to provide information on the latest research, model programs, and policy issues pertaining to this population. It describes training and technical assistance opportunities, conferences, and available resources. These resources are available in various formats, including written products, videotapes, and CDs. Many of the products can be downloaded at no cost.
http://www.uic.edu/orgs/rrtcamr/aboutus.htm

The ARC of the United States

The Arc of the United States is the national organization of and for people with mental retardation and related disabilities and their families. It is devoted to promoting and improving the quality of services for consumers and families.
The following site is dedicated to aging:
http://www.thearc.org/aging.html
A glossary of health and medical terms is available at:
http://www.thearclink.org/glossary/index.asp

UK Down's Syndrome Association

This site has available many pamphlets on different aspects of health.
http://www.downs-syndrome.org.uk/

UK Resources for Down's Syndrome

http://www.43green.freeserve.co.uk/uk_downs_syndrome/ukdsinfo.html

Alzheimer Disease

Alzheimer's Association. Down Syndrome and Alzheimer's Disease

A reviewed collection of items prepared by the Green-Field Resource Center staff.
http://www.alz.org/Resources/Resources/rtrldowns.asp

Alzheimer Society of Canada

http://www.alzheimer.ca

Alzheimer Society Ontario

As of 2006, there are 39 Alzheimer Societies in Ontario. Check this site for the one nearest you.
http://alzheimerontario.org/english/home/default.asp?s=1

Alzheimer Society Toronto

This organization has many reference materials available for loan or reference use to individuals living within the Toronto area. Individuals from outside Toronto should contact the Alzheimer chapter in their area.
http://www.alzheimertoronto.org/

Alzheimer's Australia

http://www.alzheimers.org.au/

Alzheimer's Society (U.K.)

http://www.Alzheimer's.org.uk/

Developmental Disabilities and Azheimer's Disease…What You Should Know (1995)

Developed by the *Workgroup on Alzheimer Disease and Developmental Disabilities* and the *New York Caregiver Assistance Project in Aging and Developmental Disabilities*; published by The Arc of the United States.
http://www.tharc.org/misc/alzbl/html

Development Services Development Centre Services to the Carers of People with Dementia; University of Stirling, Scotland

This site has a reading list on the topic of dementia in the intellectual disabilities that is updated regularly.
http://www-cgi.stir.ac.uk/htsearch

Intellectual Disabilities and Dementia

School of Social Welfare, University of Albany, State University of New York. A resource site for staff caring for individuals with

intellectual and other developmental disabilities who are showing signs of dementia. Administrators and researchers will also find some of the links helpful.
http://www/albany.edu/aging/IDD/

The Alzheimer's Disease Bookstore
A list of books on Alzheimer's disease by category.
http://www.alzheimersbooks.com/

The Alzheimer Society of Ireland
http://www.alzheimer.ie/

University of Illinois at Chicago
Go to the home page http://www.uic.edu/index.htm. Search for intellectual disabilities. Readers will be directed to many different topics relevant to dementia including:

- Working Resources List on Dementia Care Management and Intellectual Disability
- Resources on Alzheimer's Disease and People with Intellectual Disabilities
- Alzheimer's Disease International's Fact Sheets on Dementia and Intellectual Disabilities
- Dementia Care of Adults with Intellectual Disabilities (ID). Matthew Janicki, Philip McCallion, Lawrence T. Force, and Arthur J. Dalton.

U.S. Alzheimer's Association
http://www.alz.org/

Professional Support

Ontario Association on Developmental Disabilities (OADD)
http://www.oadd.org/

University Resources

University of Western Ontario Developmental Disabilities Division
Education Training Package in Developmental Disabilities for Medical Undergraduates in Ontario.
http://www.psychiatry.med.uwo.ca/ddp/mededucation/titlepage.htm