Appendix A

Syndromes and Diagnostic Features

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Syndromes/ Genetic Causes	Diagnostic Features	Behavioural Phenotype	Medical/Psychiatric and Medication Vulnerabilities	Psychological Vulnerabilities	Social Implications/ Vulnerabilities
Autism	Delayed patterns of social, affective, and communication development speech delay impaired social interaction restricted activities	Hyperactivity Short attention span Impulsivity Aggressivity Self-injurious behaviour Abnormal eating/ sleep patterns Sensitivity to sounds	EEG abnormalities/ seizure disorders Coexistence with other neurological medical conditions (soft neurological signs) No specific medication available Medication addresses symptoms (i.e. ,amphetamines for ADHD, antipsychotics for aggressivity, SSRI's for behavioural/anxiety depressive disorders	Period of normal development (1-2 years) Low IQ in 75% (developmental disability) Language comprehension lower than vocabulary Extremely high abilities in certain areas (music, math, reading)	Social withdrawal/ isolation Inflexibility to routines or rituals Insistence on sameness Distress to changes of routine Stereotypic body movements Abnormal focus of interest
Rett's Syndrome	Deterioration from apparently normal development in infancy or early childhood Slow down of head growth Lack of interest in the environment Loss of hand use/ stereotypic hand movements	Anxiety in response to external situations Brief and consistent screaming Hyperventilation Self-inflicting behaviour Frightened expressions and general distress Low mood/crying spells Biting of fingers and hands	Hypoplastic cold blue feet Scoliosis Changed sensitivity to pain Intensive eye communication EEG abnormalities Breath holding Bloating Bruxism Night laughing No specific medication available; only treatment of symptoms and problem areas	Severe to profound degree of developmental disability Motor and language impairment Similarities with autism and difficult to differentiate at an early age	Behavioral issues tend to be misunderstood Inability to self help skills can be easily mistaken Wetting, soiling present causes social issues Laughter and screaming can be misdiagnosed or behaviors Inability to speak is socially isolating and causes frustration

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Down Syndrome	Chromosomal disorder Trisomy 21 occurs in 94% of Down Syndrome Trisomy 21 occurs in 4% of all pregnancies 25-40 genes have currently been identified with chromosome 21 Translocation: part of chromosome transferred to another location (f. 21:13)	Appears more passive Less reactivity Lower adaptability Require more stimulation Less persistent More inflexible and resistive to change Require more attention Conduct problems	Brain weight in children less Brain abnormalities Seizure disorders (1:3 ratio) Early memory and cognitive skills loss before the age of 50 years Eye problems; keratoConus, strabismus, blepharitis Oral problems: maxilla and mandible are smaller Heart defects Lungs are smaller Gastrointestinal anomalies Skin problems Medication– treating symptoms and/or physical problems	Failure to develop normal language Central auditory processing abnormalities Expressive language difficulties is a hallmark of this syndrome Reduced short-term memory and distractibility	Social withdrawal/ passivity in some Diminished social competence in some Difficulty learning reference cues Difficulty transferring goal-directed play skills (from a joint group to individual play situations) Greater inflexibility to routines or rituals
Gilles De La Tourette Familial with genetic vulnerability Basal ganglia development is abnormal Possibly an autoimmune disorder	Chronic motor and verbal tics/ they may appear together or separately More males/females 6-8 males/1000 4-5 females/1000	Destructibility Academic/social problems Conduct problems Anxiety with rigidity of thinking and compulsion Anxiety disorder	Comorbitiy of OCD Comorbidity of ADHD Learning disorders Conduct disorders Medication neuroleptics Haldol, Pimozide, and Clonidine SSRI's stimulants for ADHD and vocal tics	Academic/learning problems Developmental delays Specific reading/math disabilities	Social impairment Behavioural problems Difficulties settling in a given task or performance Tic disorders are "stress-sensitive"

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Williams Syndrome Deletion of chromosome 7 q11.23 Multisystem disease	Dysmorphic facial features Cardiovascular disease Delayed development	Attention problems Excessive worrying Talking too much Not eating well Overfriendly to strangers Oversensitivity to sounds Negative mood	Hyperopia/strabismus Chronic otitis media Voice low pitched & coarse Aortic narrowing Hypertension GI Gastrointestinal: chronic abdominal pain/constipation Kidney/urinary problems Hyperextendible joints Hypercalcemia and failure to thrive Hypotonic initially– hypertonic later diabetes	Grammar is somewhat delayed Vocabulary is of relative strength, although developmental delays are common Onset of language delayed Auditory role memory is a strength Visual-spatial construction drawing low block design low	Performance on socialization is high/ overfriendliness/ advanced interpersonal skills Performance on communication and on daily living skills lower than other domains Anxiety disorders quite common Many can achieve independent living with regular employment Treatment of physical problems and anxiety are required
Prader Willi Syndrome	Caused by genetic alteration of the 15 chromosome (Del 15 q11-13) 1 in/15 000 Lack of paternal contribution by the specific chromosomal region	Often severe problems Temper tantrums Stubbornness Oppositionality Rigidity Crying Stealing, especially with regards to food Obsessions Underactivity Excessive sleep Anxiety	Hypotonia Obesity Hypogonadism Facial features distinct Short stature Viscous saliva High pain threshold Skin picking Osteoporosis	IQ usually around 70 Range from average to profound disability Impaired adaptive functioning Low adaptive performance with significant behavioural dysfunction and a persistent drive to eat Language development delayed	Persistent drive to eat is associated with increased weight behaviour difficulties family stress Language and coping skill delays increase risk of maladaptive behaviours Psychiatric disorders such as OCD, anxiety, and depressive disorder

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Fragile X Syndrome Mutation in fragile X gene (DNA testing identifies carriers in addition to individuals who are also affected) Most common cause of developmental disability 30% or all X-linked forms of DD	Language delay Motor delay Hypotonia Hyperactivity	Short attention span Impulsivity Hyperactivity Hypersensitivity to sounds Hyperarousal Tactile defensiveness Tantrums daily Aggressivity Transition can be very difficult Perseveration Autistic-like features	Prominent ears Long face Hyperextensibility of joints Flat feet Soft skin Cleft palate Macroorchidism Higher growth in early years Social anxiety Personality disorder Seizures (20%) Chronic otitis media Medications stimulants Clonidine Folic Acid (helpful to 50%) mood stabilizers are useful (Tegretol, Epival)	Males developmental disability sensory integration problems motor coordination impaired hyperarousal/ disinhibition Females mild degree of DD learning disabilities organizational problems math difficulties tangential speech mood lability	Hyperarousal Sensitivity to sounds and other stimuli Avoidance of crowds Social isolation Behaviour and personality difficulties Poor eye contact Easily misdiagnosed as Autism
Phenylketonuria Autosomal recessive Inherited from both parents Affect both males and females in equal numbers	If untreated: phenylalanine toxicity dopamine reduction developmental disability eczema seizures ataxia motor deficits behavioural problems	Newborn screening prevents the disorder If missed: developmental delay delayed milestones When treated even later, many symptoms disappear ADHD OCD Autistic like features	Treatment phenylalanine restricted diet (special formula) diet discontinuation before 10 yrs of age creates delays in development discontinuation after 10 yrs. may be tolerated, but subtle psychological consequences exist Personality and temperament disturbance Medication stimulants not very good	Visual-motor deficits are prevalent Fine motor speed is diminished Problems in mental processing Executive functioning (ability to retain information and use it later for problem-solving) is diminished When “cognitive load” is increased, children become confused	Factors influencing outcome: age at initial treatment lifetime level of metabolic control current dietary status Psychiatric and learning comorbidity causes social problems and behavioural issues Agoraphobia/depression common in adolescence/ adulthood

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Seizure Disorder Due to inherited or acquired neurodevelopmental abnormalities Sudden discharge of electrical activity in the brain that results in alteration of sensation, behaviour, or consciousness	Epilepsy is condition of recurrent seizures Epileptic syndrome is characterized by a recurrence of consistent symptoms and behavioural manifestations	Impairment of attention/ concentration Ability to learn and remember material is impaired Decreased reaction time and decreased psychomotor speed Impairment and exaggeration of sensory input	Sensory misperceptions/ hallucinations Objects are perceived as smaller (micropsia) Visual misperceptions and auras- usually olfactory or gustatory Abdominal and epigastric sensations Cephalgic auras and as sharp pain on the head easily misdiagnosed as migraines The above features are experienced by these persons as dissociations and not as real events Medication antiepileptics anticonvulsants	Disturbances of executive functions Sensory systems are affected Motor systems are affected Language skills and reading acquisitions are affected Dysnomia and anomia are linked to complex partial seizures and are easily misdiagnosed as memory problems Hypergraphia is common Circumstantiality of written and spoken language is evident	Specific personality characteristics have been associated with various forms of seizure disorders hyperactivity/inattention rigidity obsessive compulsive features developmental delay specific learning disabilities All of the above features can be misinterpreted as laziness, stubbornness, and eccentricities