Demystifying Syndromes: 
Behavioural Implications of 
Genetic Syndromes

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• Why it is important to have a correct diagnosis 
• Fragile x Asperger Disorder 
• 22q11.2 deletion Down Syndrome 
• Smith Magenis Fetal Alcohol Syndrome 
• Williams 
• Angelman 
• Prader-Willi

Approximately 750 genetic disorders associated with 
developmental disability 
Can find a genetic etiology in 60 % of individuals with 
developmental disabilities (Finucane, 2005)
What is a behavioural phenotype?

The behavioural phenotype is a characteristic pattern of motor, cognitive, linguistic and social abnormalities which is associated with a biological disorder. In some cases, the behavioural phenotype may constitute a psychiatric disorder; in others, behaviours which are not usually regarded as symptoms of psychiatric disorders may occur.

Flint & Yule, 1994

Cautions (Brenda Finucane, 2005)

- Don’t accept behaviour as inevitable and therefore do nothing about it
- Don’t accept behaviour as inevitable and therefore contribute to its occurrence and maintenance

- Currently access to services is driven by educational or psychiatric diagnoses
- Developmental Disability as a symptom
- Psychiatric diagnoses are lists of symptoms
- What is underlying cause?
- Etiological causes often considered unimportant in managing behaviour
Diagnostic Alphabet Soup

Why is etiology important?
- Syndrome driven behaviour
- Specific learning styles
- Medical needs often specific to a syndrome
- Genetic counseling for family
- Alleviate guilt
- Correct misconceptions
- Support for families
- Research

Genetics: the basics
- Chromosome
  - Extra chromosome
  - Translocation/Duplication/Insertion/Inversion
  - Deletion
  - Mosaicism
- Metabolic Disorders
- Testing
  - Blood analysis
  - FISH (Fluorescent in situ hybridization)
  - DNA analysis
**Importance of Family History**

- Depression
- Tourette S
- ADHD, Tics
- Histrionic PD
- Asperger
- Dev. Dis.
- ?
- Asperger
- Autism

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**Fragile X**

- 1/2500 males in general population
- In Developmental Disability may be as many as 1/25 males
- Females also affected – more variable symptoms
- Under-diagnosed
- Most common familial cause of Dev.Disability
- Distinctive cognitive and behavioural profile

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**FrX-Classic facial features**

- Long face
- Prominent chin and forehead
- Prominent ears with cupping
- Dental crowding
Fragile X – Common Behavioural Characteristics

- Gaze aversion
- Resistance to change
- Social anxiety /shy
- Distractibility /hyperactivity
- Hypersensitive to sensory stimuli
- Behavioural difficulties related to anxiety or hyperarousal
- Up to 1/3 of those diagnosed with autism also have fragile x (National Fragile X foundation)

Gaze Aversion

The greeting behaviour of fragile x males. AJMR, 93, 408 (1989)

FrX-Medical vulnerabilities

- Cardiac problems –
  - mitral valve prolapse
  - Dilation of the aorta
- Middle ear infections
- Sinus infections
- Abnormalities in eyes
- Seizures (20%)
FrX-Cognitive Strengths
- Simultaneous processing of visual information
- Imitation
- Excellent long-term memory
- Respond well to structure and routine

FrX-Cognitive weaknesses
- Poor auditory memory
- Poor sequential processing
- Weak abstract reasoning
- Difficulty with novel tasks
- Poor visual motor integration

22q11.2 Deletion Syndrome
- 22qDS
- DiGeorge Syndrome
- Velocardiofacial Syndrome
- Shprintzen Syndrome
- Conotruncal Anomaly Face Syndrome
- Opitz G/BBB syndrome
22qDS
- 1/2000 to 1/5000 births
- Under-diagnosed; testing began in late 90's
- Use FISH to diagnose
- Common cause of learning and developmental disabilities
- 10% inherited from parent
- Highly variable physical and cognitive effects

22qDS-Medical vulnerabilities
- 80% born with cardiac defects (Finucane 2005)
  - range from minor to severe
- Palate abnormalities
- Hearing impairment
- Structural kidney anomalies
- Endocrine problems
- Autoimmune disorders
- Scoliosis
  - ***Almost any organ can be affected***
22qDS-Cognitive Weaknesses

- Just beginning to understand the syndrome
- Delayed motor and speech development
- IQ ranges from average to moderate DD
  - Approx. 40% have developmental disability
- Poor nonverbal processing
  - May be diagnosed as nonverbal LD
- Poor visual spatial abilities
- Poor attention, working memory and executive functioning
- Poor visual memory – esp. for faces
- Poor math skills

22qDS-Cognitive strengths

- Higher verbal IQ than nonverbal
- Rote verbal learning and memory
- Auditory perception and memory
- Relatively good word reading and decoding
  - However, poor reading comprehension

22qDS-Emotional/behavioural problems - children

- ADHD
- Temper/emotional outbursts
- Poor impulse control
- Social problems
- Anxiety
- 1/3 – ¼ meet the DSM criteria for an Autism Spectrum Disorder in childhood (Finucane, 2005)
22qDS - Emotional/Behavioural problems – adolescents/adults

- Higher frequency of psychiatric disorders (Boyd et al., 2004)
  - 36% of adults with 22qDS had a psychiatric history; many psychotic
  - 64% had some kind of diagnosis
  - 1/50 schizophrenics has 22qDS (Finucane, 2005)
  - 20-25% meet the DSM criteria for schizophrenia or schizoaffective disorder

Smith-Magenis Syndrome (SMS)

- Deletion on one of the number 17 chromosomes
- Generally not inherited
- Usually mild to moderate Dev. disability
- 1/25,000 live births
  - But, widely considered to be under-diagnosed
  - In a population of 1000 individuals with Developmental Disabilities, 23 were diagnosed with SMS (Finucane & Simon, 1999)

Smith-Magenis

- Two stereotypic behaviours
  - Self hug
  - “Lick and flip”
- Sleep disorder – chronic
  - Abnormal circadian rhythm of melatonin
Self Injury
- head banging
- Orifice stuffing
- Fingernail pulling
- Decreased sensitivity to pain
- Peripheral neuropathies
- Visual
  - Strabismus, nearsightedness
  - Retinal detachment

Hearing deficits
- Otitis media
- Cardiac malformations
- Kidney malformations
- Sleep disorder

***If SMS goes undiagnosed, medical conditions may be undiagnosed resulting in permanent damage.
**SMS-Positive characteristics**

- Engaging, endearing, full of personality
- Like attention, eager to please, helpful
- Good sense of humour
- Communicative
- Responsive to structure and routine
- Respond to reinforcement
- Can redirect if caregivers are alert
- Visual learners
- Good long term memory

**SMS-Challenges!!!**

- Caregiver sleep deprivation
- Attention seeking
- Perseveration of questions
- Aggressive hugging of others
- Poor impulse control
- Prolonged outbursts
- Poor sense of time
- Difficulty waiting
- Need to keep active
  - Short but high-interest activities
- Emotional toddlerhood (Finucane, 2005)
  - Easily frustrated
  - Emotionally volatile
  - Temper tantrums
  - Reactive

**Supports for SMS**

- Melatonin – with med holidays
- Physical and neurological examination
- Structure and routine
- Remain emotionally neutral
- Avoid power struggles
- Have a large “bag of tricks”
- Prevention – recognize early signs
- Redirect
Williams Syndrome

- Characteristic appearance
- 1/20,000 births
  - 1/200 in individuals with Dev. Disabilities
- Most in mild to moderate range of DD
- Deletion on chromosome 7
- Distinctive personality
- Presents higher than ability level

http://www.Williams-syndrome.org/

WS – Medical vulnerabilities

- Cardiovascular abnormalities (80%)
- Vision problems
- Chronic ear infections
- Increased sensitivity to sound (90%)
- Hypertension: adults and children (50%)
- Hypercalcemia as infant (15%)
- Urinary tract infection
- Renal difficulties (18%)

WS- Strengths

- Well developed expressive language
- Friendly, sociable, charming, loving, caring
- Musical talents
- Good facial recognition and memory
- Good emotional recognition, empathetic
- Good short-term memory

***talk through problems / verbal mediation for problem solving
WS – Challenges

- Overly friendly – no stranger fear
  - Major impairments in visual-spatial tasks
- High rates of anxiety, fears
- Poor attention
- Unusually sensitive to noise
- Poor motor control
- Poor concept of time/money/math skills

Angelman Syndrome

- “happy puppet syndrome”
- Severe developmental disability
- Nonverbal
- Late to walk – ataxic gait
- Happy demeanour frequent laughing
- Seizure disorder
- Often diagnosed as CP

AS - Universal Features (100%)

- Severe developmental disability
- Movement or balance disorder – ataxic gait
- Frequent smiling, laughing
- Absent or minimal expressive speech
- Relatively better receptive language
- Hyperactivity
- Short attention span
- Adaptive behaviour higher than IQ predicts
AS – Frequent features

- Sleep disorder – night wandering
- Seizure disorder
- Hand flapping when excited or walking
- Chewing, mouthing behaviours
- Grabbing, pinching, pulling hair
- Fascination with water, shiny objects

Guidelines for support of AS

- Constant supervision!!!
  - Adaptations to environment
  - Behaviour is communication
  - Redirect activity
  - Bartering works!
  - Consistency - routines
  - Remember they can understand you!

Prader-Willi Syndrome

- Usually associated with a deletion on paternal copy of chromosome 15
- 1/20,000
- Physical characteristics:
  - Short stature
  - Small hands/feet
  - Underdeveloped secondary sex characteristics
  - Facial appearance
- Leading known cause of obesity
- Food seeking, hoarding
- Compulsive behaviours
- Good visual spatial skills (puzzles)
PWS: Medical Vulnerabilities
- Sleep disturbance, sleep apnea
- Health related problems associated with obesity
- Hypotonia, poor muscle tone
- High pain threshold
- Diabetes
- Osteoporosis

PWS – Cognitive factors
- Relatively high IQ
- Specific strength in visual patterning
- Visual processing
- Simultaneous processing
- Short-term memory deficits
- Adaptive behaviour less than predicted by IQ

PWS Behaviour
- Stubborn, impulsive
- Lying, Stealing, hoarding
- OCD tendencies
  - Perseverating
  - Need to put things in order
  - Repetitive actions
  - Skin picking
- Tantrums and Violent Outbursts
- Depression is not uncommon
- High pain threshold may lead to behavioural issues
PWS: Weight control

- 1,000 – 1,200 calories/day
- Adequate calcium intake
- Regular exercise
- Environmental controls
- Close supervision, behavioural approach

Guidelines for Support - PWS

- Need more supervision than IQ suggests
- Behavioural issues are Life Long, therefore strategies need to be sustainable.
- External structure in all areas related to food
- External structure related to money and telephone access.
- Consistency, limit setting, redirection
- Cue for transitions
- Avoid power/control issues.

Down Syndrome: Basic Facts

- 1:800 births (some say 1:600)
- Abnormality in 21st chromosome
  - 92% Extra 21
  - 2-3% Mosaic
  - 3-5% unbalanced translocation/other abnormalities (Dykens, Hodapp & Fincane, 2000)
- Usually not familial
DS: Basic Facts 2

- Characteristic facial features
- Occurrence increases with maternal age
  - BUT more than 80% are born to women younger than 35
- Developmental delay / disability
- Average life expectancy is 55 years
A Down Syndrome Advantage?

- Widely recognized
  - Support groups
  - Social orientation
  - Perceived as cheerful and lovable
  - “baby-face overgeneralization” (Dykens, Hodapp & Finucane, 2000)

Intellectual ability

- can not predict future development from diagnosis alone
- range of intellectual ability
  - Average IQ is 55
  - 10% in “normal” range (average to low average) (Griffiths & King, 2004)
  - likely to have learning disabilities
- 10% profound and multiple disabilities
- development does not plateau in adolescence

Medical Complications

- Hearing deficiency (75%)
  - children to age 13 reassess hearing yearly
  - teens and adults reassess hearing every 2 years
  - otitis media (ear infections) 50 - 70%
- disorders of vision (60%)
  - assess vision yearly to early teens, thereafter every two years
Medical Complications cont.
- Congenital heart disease (50%)
  - wide variation in type and severity
- Sleep-related problems
- Disorders of thyroid function (15%)
- Cervical spine abnormality (10%)
- Obesity - resting metabolic rate lower
- Cavities - periodontal disease

Language
- Delayed speech and language development
- Receptive language skills develop faster than ability to communicate
- Specific impairment in short-term memory for verbal information
- Difficulty processing verbal information
  - therefore difficulty learning through listening

Language cont.
- Particular impairment in grammatical ability
- Articulation difficulties
- Hearing and oral structure problems contribute to language difficulties
Learning abilities

- Visual-spatial abilities stronger than verbal abilities
- Relative strength in the ability to learn from visual information
- Delays in motor abilities
  - Speech-motor difficulties
  - Gross and fine motor delays

Learning Abilities

- Research just beginning into development of reading, writing and numeracy skills (most research in UK)
  - Steady progress through school and into adulthood
  - Reading a relative strength
  - Numeracy skills often weaker than reading ability

Learning abilities cont.

- UK Research
  - Children in mainstream programs have significantly better language and literacy skills than their same ability peers in segregated classes (special schools)
  - Also better numeracy skills
  - General social and independence skills did not differ
  - Interpersonal friendship skills better in children from "special schools"
Social Development

- Social emotional development a strength in both children and adults
- 2/3 no major behaviour problems
- Range of personality and temperament same as in normal population

Is there a Down syndrome Personality?

- Gibson (1978) noted the stereotype included five aspects: amiable nature, some obstinacy, keen sense of the ridiculous, imitation, and excellent memory
- “Sociability”

0 - 5 years

- Development follows same pattern as normal children but at slower rate
- Progress influenced by stimulation and love
- By five most walking and toilet trained
- Communication likely delayed
- Age-appropriate placement in school
5 - 11 years
- Progress in speech and language skills
- Developing personal self-help skills and personal independence
- Develop social/interpersonal skills
- Expect age-appropriate and socially accepted behaviour
- ADHD?

Teens
- Puberty same age as normal teens
- Typical leisure interests
- Social and emotional needs are age-appropriate
- Personal identity development
- May have some frustration at not being able to participate to same extent as normal peers
- Typical awareness of sexuality
Late Teen to Adulthood
- Awareness of sexuality
- Development of relationships and friendships
- Continue to develop appropriate social skills
- Relative independence
- Employment

Sexuality
- Dating normal part of adolescence and adulthood
- Personal safety important
- Fertility rates lower in women, but can still get pregnant!

Middle years onwards
- Menopause often 10 years earlier than normal population
- Monitor behaviour changes/loss of skills
- Recommend exercise and nutrition
Psychiatric Disorders (or not)

- Incidence of psychiatric disorders same as in normal population
- Behaviour sometimes mistaken for a psychiatric disorder
  - Self-talk
  - Imaginary friends
  - 81% adults engage in conversations with themselves or imaginary companions
    - (McGuire, Chicoine & Greenbaum, 1997)

Depression in DS

- Tendency for depression in older adults
- 10% suffer from affective disorders
  - However, in DS diagnosed significantly less often
  - May be confused with dementia
- Bipolar rarely seen

OCD in DS

- OCD features – often more ritual/routine
  - The “Groove”
- OCD is an anxiety disorder
- Association between traumatic experiences and onset of OCD behaviour
  - (Bowerkeki & Antoci2004)
- Compulsive Behaviour Checklist
  - Ordering, completeness, deviant grooming, cleaning, checking/touching
DS and Alzheimer Disease
- Greater risk?
- Chromosome 21 linked to AD
- Almost all DS will have AD pathology by age of 40
- BUT not all will develop dementia

Estimated rates of AD in DS
- Estimated rates of AD by age group
  - 40-49 10-25%
  - 50-59 20-55%
  - 60-69 30-75%

DS and Aging/Dementia
- Dementia means that someone has lost cognitive skills – it is not necessarily a disease
- There are MANY possible causes for dementia – some are reversible (e.g., depression, B12 deficiency)
- Need full medical to rule out medical reasons for changes
Medical Causes or AD?

- Vision and hearing
- CBC with differential blood chemistry
- Thyroid function
- Vitamin B12
- Folate (folic acid)
- medication review
- CT/MRI

Mood disorders or dementia?

- Depression: common symptoms to both DS and AD
  - Loss of adaptive skills, Memory loss
  - Disruption of sleep cycle, appetite changes
  - Apathy, moodiness, irritation
  - Psychomotor agitation or retardation
  - Presence of psychotic features
- Depression may coexist with AD
- Symptom course may differentiate between AD and DS

Criteria for probable diagnosis of AD

- Progressive decline in function
- Progressive memory loss
- Gait apraxia
- Incontinence
- Seizures
- BUT these alone are not sufficient to make the diagnosis unless all other possible causes have been ruled out
**Diagnosis**
- Medical testing to rule out physical causes
- Evaluation of life circumstances and environment
- Baseline psychological assessment
- Dementia Rating Scale for Down Syndrome
- Follow-up at 6 to 12 months

**Early signs of AD**
- Depends on premorbid cognitive level
- In mild/high moderate ability group
  - Intellectual signs first
- In Moderate to severe ability group
  - Behavioural signs first
  - Seizures

**Care management of AD**
- Safety
- Stability of environment
- Social emotional issues
- Ongoing health monitoring
Resources