Pervasive Developmental Disorders

also known as Autism Spectrum Disorders

Dr. Deborah Marks
Pervasive Developmental Disorders

- Autistic Disorder (Autism) - Kanner
- Asperger Syndrome
- Pervasive Developmental Disorder-Not Otherwise Specified
- Rett Syndrome
- Childhood Disintegrative Disorder
Behavioural Definition: Triad of Impairments

- social interaction
- social communication
- symbolic thinking / creative play
- SPECTRUM/CONTINUUM OF SYMPTOMS presents differently according to age, gender and IQ
- Symptoms present by 3yrs: later presentation if better language skills
Diagnostic Process

- Multi-disciplinary team together or in sequence
  - Paediatrician: Diagnosis; symptoms on history, medical conditions, family situation, planning for future
  - Speech Pathologist: functional and formal language skills
  - Psychologist: developmental level/IQ and style of thinking and interaction, adaptive skills
  - Occupational therapist: play skills
Criteria for Autistic Disorder
-DSM-IV

- Qualitative impairment of social interaction (at least 2)
  - impairment in non-verbal behaviour such as eye contact
  - failure to develop appropriate peer relationships
  - lack of spontaneous seeking to share enjoyment and interests
  - lack of social or emotional reciprocity
Criteria for Autistic Disorder - DSM-IV

- Qualitative impairment of Communication (at least 1)
  - delay in spoken language without non-verbal communication
  - inability to sustain conversation
  - lack of social imitative or make-believe play
  - stereotyped, repetitive or idiosyncratic language
Criteria for Autistic Disorder - DSM-IV

- Restricted and Repetitive Patterns of Behaviour, Interests and Activities (at least 1)
  - preoccupations with patterns of interest of abnormal intensity or focus
  - inflexible adherence to non-functional routines
  - stereotyped and repetitive motor mannerisms
  - persistent pre-occupation with parts of objects
Asperger Syndrome

- Same criteria for social impairment and for repetitive and behaviour and restricted interests
- No clinically significant language delay defined as sentences + verb by 3 years (disputed)
- No cognitive delay
- Normal self-help skills and curiosity
- Often present late
- Difference between AS and high functioning autism debated
Pervasive Developmental Disorder-Not Otherwise Specified

- poorly defined group that do not meet criteria for autism or Asperger syndrome but have symptoms and thinking style of children with PDD
- alternative often given is Autism Spectrum Disorder
Spectrum of Conditions

- Pervasive Development Disorder - NOS
- Classic Autism
- Classic Asperger Syndrome
- Kanner
Early Indicators of ASD

- Lack of social smile and responsive facial expression
- Limited social language/babble
- Preference for solitude: lack of eye contact and social interest
- Lack of pointing to items of interest (not needs)
- Sensory hypersensitivity/hyposensitivity

Potential screening with M-CHAT
Other Key/Common Features

- Overfocusing
- Difficulties retrieving relevant information in spite of good memory for detail
- Poor understanding of underlying concepts
- Motor co-ordination and planning problems
- High anxiety levels
- Regression at 18months – 2 years: 20-25%
- Unusual sensory reactions
Perceptual Abnormalities

- Sound sensitivity
  - Generally for machine noise, human voices
- Visual ‘obsessions’
  - Staring into space or at water
  - Peripheral vision
- Tactile defensiveness
  - Haircuts and clothing textures
- Eating disorders
  - Hypersensitivity to flavours and textures
- Hyposensitivity to pain
Savant skills

- Present in approx 10%
- Hyperlexia
- Calendar calculation
- Perspective drawing
- Numerical calculation
Differential Diagnosis

- **Language disorders**: social functioning and understanding of others is preserved
- **Intellectual disability**, especially moderate ID
- **Selective mutism and social avoidance disorders**: lack repetitive behaviours
- **Reactive attachment disorder**: improves with change in environment
- **Tourette’s Syndrome**
- **Attention Deficit Hyperactivity Disorder**
Associated Conditions

- Intellectual Disability
  - Estimates vary from 70% to 30%
- ADHD, Anxiety Disorders, Mood Disorders – 70%
- Macrocephaly 20%
- Learning difficulties - common
- Hearing &/or Visual Impairment
- Epilepsy 17%
Proposed mechanisms

- Systematizing end of Systematizing/empathizing spectrum (previously known as ↓ theory of mind)
- Central Coherence (ability to extract relevant information) is weak
- Executive functions, including planning and impulse control, are deficient.
Pathology

- Disorder of neuronal connectivity
  - Spread of excitation between cortical minicolumns
- Association of MECP2 (Retts Syndrome) and Fragile X with autistic features supports synaptic dysfunction
- Accelerated head growth occurs at time of regression/symptom development
- FMRI implicates mirror neuron deficit and facial recognition areas.
Causes

- Most are genetic: 90% identical twin concordance rates
- Known genetic syndromes present in minority of children ~10%
  - Fragile X
  - Tuberose Sclerosis
  - duplications 15q1-q13; 16p11
- Intrauterine causes: Valproate (PKU)
Treatment

- Evidence supports educational interventions
  - Joint attention, communication, and social skills
  - Applied Behaviour Analysis: discrete trial training
  - Treatments centre-based or involve parents e.g. TEEACH
- Uncertainty about intensity needed, age of onset
Education and Early Intervention

- Focus on joint attention, communication and social skill development
- Generalist and Specialist types
- Special education settings
  - autism specific special schools
  - special schools
  - mainstream schools with integration aide
General Support Measures

- Ongoing parent support work, deal with parental depression and anxiety
- Genetic counselling
- Individual cognitive behaviour therapy sometimes effective for Asperger Syndrome
- Respite
- Family support groups
- Financial assistance
- Special support at times of transition
Long Term Needs

- Specific training in social skills: supportive therapy in social development
- Specialized employment and training facilities and supports
- Specific psychological support in sexual development
- Treatment of co-morbid conditions such as anxiety disorders
Genetic Counselling

- Greater risk of subsequent children with autism:
  - 8% if male, 2% if female
  - ~25% if have 2 or more sibs with ASD
- 20-50x risk of recurrence in subsequent pregnancy
Drug Therapy

- NO TREATMENT FOR CORE SYMPTOMS - use for comorbid symptoms such as aggression, mood swings, self-injury
- ‘Treat’ environment first
- Use multidisciplinary approaches
- Use lowest possible doses
Drug Therapy

- **Risperidone** preferred for aggression - side effects still important, recent PBS listing
- **SSRIs** useful for anxiety, depressive and certain rigid thinking patterns
  - Restlessness, agitation and insomnia may limit use
Drug Therapy

- **Stimulants** - Ritalin
  - reduces hyperactivity, decreases impulsivity
- 50% side effects:
  - agitation, irritability, aggressiveness, sleeplessness, increased stereotypies