

Plenary Session

• Signs of Autism Spectrum Disorders in Infancy and Childhood

Aetiology

- Genetics , inherited and chromosomal abnormalities
- In Utero, disruption to the development of the temporal and frontal lobes
- Birth, unfavourable obstetric events, being premature
- Inborn errors of metabolism
- Infections of the brain in infancy

Increase in Children Diagnosed with ASD

- Currently estimated as one child in 100
- In part attributable to:
- More younger children being diagnosed (Parner et al, Danish study) A California study (Hertz-Picciotto and Delwiche) suggests this explains 12% of the increase

Increase in Children Diagnosed with ASD

- Inclusion of milder cases explains 56% of the increase (Hertz-Picciotto and Delwiche)
- Fewer children diagnosed with mental retardation accounts for 26% of the increase (King and Bearman; Nassar et al)
- *About 5-10% remains unexplained*

3 Developmental Trajectories

- 1) No period of typical development- autism signs present from birth
- 2) Developmental plateau- child achieves typical developmental milestones and then the rate of progress declines
- 3) Regression- child loses skills (20-35%)

Autism apparent in early Infancy

- The child does not seem to 'bond' with his or her mother.
- When distressed, not easily comforted by affection
- Lack of interest in social activities.
- Content to be alone.
- Suspected deafness.
- Lack of response when name called.

Autism apparent in early Infancy

- Lack of vocalizations.
- Unusual mannerisms to express emotions (especially when excited).
- More frequent and intense distress reactions
- Inexplicable crying
- Less likely to look at a parent to seek reassurance and approval

Recognised Almost From Birth

- Hyperacusis (Vacuum cleaner, sudden noises).
- Delayed reflexes.
- Hypotonia.
- Slowed motor development
- Sleep disturbance.
- Feeding problems (reflux).
- Ear infections

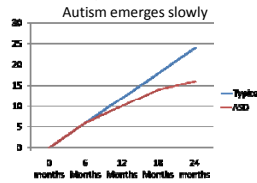
Recognised Almost From Birth

- Macrocephaly – large head circumference (25%)
- Longer or taller

Medical Signs

- Epilepsy (complex partial seizures).
- Neuro-cutaneous disorders (tuberous sclerosis).
- Chromosomal disorders (Fragile X, 2-3%).
- Candidate genes on chromosomes 2q,7q,16p,19p and 22.
- Cytogenetic abnormalities on chromosome 15.

Developmental Plateau



6 Months

- Marginally less likely to look at and seek other people
- Marginally less likely to smile and vocalize at others.
- Observations at 6 months not very predictive

At 12 Months

- Less likely to respond when his or her name called.
- Fewer gestures than typical children.
- Understands fewer phrases
- Difficulties in disengagement of visual attention.
- Look at others less frequently.
- Looking at a person's mouth rather than eyes

At 12 Months

- By 12 months, differences on almost every scale of a developmental assessment of 2-3 months.
- A global developmental delay but just within the normal range
- Macrocephaly in 25%

12-24 Months

- Diminished eye contact
- Diminished social engagement
- Limited interest in social games and turn taking exchanges
- Preference for being alone
- Visual attention more frequently to objects than people

12-24 Months

- Limited range of facial expression
- Less sharing of affect (smiling and looking at others)
- Unusual hand and finger mannerisms can emerge
- Walking on tiptoes
- Difficulty adapting to new situations and coping with changes in routine

12-24 Months

- Interest in visually repetitive phenomenon, such as ceiling fans
- Unusual attachment to an object
- Communicative acts are disproportionately imperative- 'I want that!' than declarative- 'Look at that!'
- Less likely to indicate 'Look at me' and bring an object to an adult for joint interest

12-24 Months

- Unusual sensory interests (spinning or sniffing objects)
- Afraid of some everyday sounds
- Eats a very limited range of foods
- Overly reactive to tickling

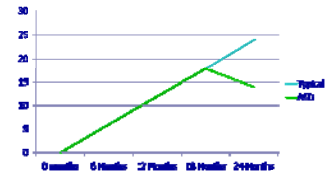
12-24 Months

- Limited imitative play
- Overactive and uncooperative
- Lack of interest in other children

12-24 Months

- Extremes of temperament
- Less imaginative or pretend play
- Less complex babbling and word production
- Delay in receptive and expressive language

Regression



Regression

- Between 10 and 30 months
- Peak at 18 months
- Loss of previously acquired words or stops talking
- Significantly less eye contact
- Lose interest in social games and being with people and parents

Regression

- Stereotyped behaviours (hand flapping)
- More irritable or anxious
- No environmental, psychological or medical explanation

Regression in ASD
Stefantos

- Occurs in 15-47% of children diagnosed with autism
- Can occur from 15-30 months, usually 18-24 months
- The regression occurs over weeks to months but can be days.
- Can be an increased head circumference (also occurs in no regression autism)

Regression in ASD
Stefantos

- 20-40% lose all expressive language
- Rarely affects self-care and motor skills
- Most have relatively minor problems prior to regression

Regression in ASD
Stefantos

- Slow recovery of language with periodic setbacks
- Regression of language unique to ASD
- May be no difference in long-term outcome in comparison to the non-regressive pathway to autism
- May be associated with a family history of auto-immune disorders

Excellent review article

- Yirmiya, N & Charman, T. (2010). The prodrome of autism: early behavioural and biological signs, regression, peri- and post-natal development and genetics. *J Child Psychol. Psychiat.*, 51(4), 432-458.

Diagnosis Statistics for autism

- Average age of diagnosis
 - Autism (AD) - 3.1 yrs
 - PDDNOS - 3.9 yrs
 (Mendell et al, 2005; US data)

Medical Exam

- Check hearing and vision
- IF INDICATED: check for chromosomal abnormalities, neuro-cutaneous disorders
- Petit mal seizures (esp. around 3 years)

Child Observation and Play

- Achieving a rapport with the child
 - Examiner to be calm and happy
 - No sudden movements
 - Voice clear but not loud
 - Eye contact not intense
 - Allow personal space



Achieving a rapport with the child

- Simple directions
- Appreciate compliance
- Avoid demonstrations of affection
- Motivation- information from parents: freedom, rough and tumble play, food.



Achieving a rapport with the child

- Remove distractions such as mirrors, toys that can spin
- Reduce background noise such as an air conditioning unit
- Check lighting (avoid fluorescent lighting)
- Gain momentum, quickly completing and succeeding at specific tasks.

Play with child

- Observe and play:
 - Reciprocity and nonverbal language
 - Expressive and receptive language
 - Type of play and play style
 - Motor skills
 - Sensory behaviours
 - Interaction with parents

Look out for:

- Self-stimulatory behaviours such as hand mannerism, rocking and mouthing
- Holding objects as a 'security blanket'
- Complex mannerisms
- Anxiety
- Aggression
- Overactivity
- Functional and symbolic use of objects

Repetitive behaviour in typical development

- Motor stereotypies (eg banging, waving, bouncing, rocking) are typical in first 12 months
- 'just right' behaviours, eg lining up objects, and insistence on sameness – typical for toddlers and pre-schoolers
- Tend to grow out of these steadily from age 3-6 years old

Video Examples of Young Children with Autism

- www.autismspeaks.org
- ASD Video Glossary
- Over 100 video clips
- Compare typical and ASD behaviours and abilities

Sub Threshold ASD

- In DSM, PDDNOS
- In ICD, Atypical autism
- Fragments, but not the full picture
- Metaphor of the diagnostic puzzle



Differential Diagnosis

- Typical development
- Global developmental delay
- Language delay/deviance
- Quasi Autism from extreme neglect
- OCD

Differential Diagnosis

- Another anxiety disorder
- Reactive attachment disorder
- Complex PTSD
- Selective mutism
- ADHD
- Rett Syndrome
- Childhood Disintegrative Disorder (normal development up to at least 2 years)

Considerations in conferring an early diagnosis

- Enormous amount of natural development that occurs between ages 3-6 years old
- False positives
- Changes in clinical profile due to early intervention
- Diagnostic stability if the child is less than 30 months – 52%
- Diagnostic stability if the child is more than 30 months – 87%

Strong genetic component

- At least 15% of families will have a second child with ASD
- 40% of siblings have some concerns with development, usually language delay, ADD.
- Broader Autism Phenotype (A parent may have a 'ghosting' of the characteristics)
- In some cases there is no evidence or indication of a genetic aetiology

Explaining the Diagnosis to Parents

- Characteristics observed in the child
- Qualities in abilities and personality
- How the child copes with having autism
- Services, resources and Early Intervention
- Range of outcomes
- Report to include strengths and weaknesses and recommendations for treatment

Indicators of a Relatively Good Prognosis

- Development of speech
- Profile of cognitive abilities
- Parental determination
- Personality of the child
- Access to effective early intervention, 20-25 hours a week.

Evaluation Studies

- Sofronoff, K *et al* (2005) 'A randomised controlled trial of a CBT intervention for anxiety in children with Asperger syndrome.' *Journal of Child Psychology and Psychiatry* 46, 1143-1151.
- Sofronoff, K *et al* (2007) 'A randomized controlled trial of a CBT intervention for anger management in children diagnosed with Asperger syndrome.' *Journal of Autism and Developmental Disorders* 37, (1203-1214).