Childhood Developmental Diversions

Juan Garcia
Catherine Swan
Jenny Gibbs
Neurodevelopmental Disorders

- Overview of neurodevelopmental disorders
  - Intellectual Disability
  - Communication Disorders
  - Autism Spectrum Disorders
  - Attention Deficit Hyperactivity
  - Specific Learning Disorder
  - Developmental Coordination Disorders
  - Stereotypic Movement Disorder
  - Tic Disorders
Overview neurodevelopmental disorders

- Group of conditions with onset in the developmental period
- Typically manifest early (often before school age)
- Result in developmental deficits that produce impairments of personal, social, academic or occupational functioning
- Varies from specific to global.
- Neurodevelopmental disorders frequently cooccur e.g. ID, ASD; ADHD, SLD
- Not just childhood conditions; have lifelong impact
• Neurodevelopmental disorders frequently overlap
• Deficits are in a continuum with the normal range
• Polygenetic
Overview of workshop

• Developmental Delay and ASD – Catherine
• School age children with learning difficulties - Jenny
  – Intellectual Disability
  – Specific Learning Difficulties
  – Special Education in NZ
• Dyspraxia/Developmental Coordination Disorder - Catherine
• ADHD and its management – Juan
• Questions and discussion
Global Developmental Delay

Catherine Swan
Developmental Paediatrician CDHB
...where we all began...

Modifying factors

– Genetic
  • Inherited and sporadic

– Embryonic
  • Toxins
  • Nutrition
  • Process problem

– Fetal
  • Infection
  • Trauma
  • Stress
  • Nutrition
  • Vascular

– Delivery
  • Timing
  • Infection
  • Trauma
  • Complications
Child development

DIR model - Greenspan et al

Biologically based individual differences

Family, community, cultural

Child-caregiver interactions

Development
Global developmental delay

- Significant functional delays in 2+ areas of development
  - motor (gross/fine)
  - speech/language (expressive, receptive, mixed)
  - cognition
  - Social/play
  - Functional

- Under 5 years (DSM 5)
- Up to 8 years (NASC)
- 2 standard deviations from mean
Delay versus disorder

- GDD is generally a misnomer
- Implies “catch up”
- For majority gap will widen with age
- Aim to further define between 4-8 years
  – For most diagnosis will be Intellectual disability
Facts and figures

• 2-3% of the population
• 2/3 mild-to-moderate level of impairment
• 1/3 severe-to-profound level of impairment.
Recognising and referring
Recognising

• Parental concern ("She’s not like the others")
• Wellchild – PEDS
• Supplementary info
  – Queensland milestones
  – Denver (as way to describe; not as screening tool)

• Parental concern as good/better predictor vs Health professional
Other parts of developmental history

– Birth history
– Home
– Family
– Health
– Vision/hearing
Red Flags – Social and Language

All children with ANY of the following findings should be referred for a further assessment:
• no babble, pointing to or showing of objects or other gesture by 12 months
• no meaningful single words by 18 months
• no two-word spontaneous (non-echoed or imitated) phrases by 24 months
• ANY loss of any language or social skills at ANY age.
Red Flags - Motor

• Asymmetrical movement
• Not sitting by 10 months
• No means of mobility at 12 months
• Not walking at 18 months
• Seems stiff or floppy or loose
• Difficulty grasping manipulating objects
Workup?
Karyotype

• Karyotypes describe the number of chromosomes, and what they look like under a light microscope.

• Limitations – extra or missing pieces have to be visible under light microscope.
Microarrays

Group of complex technologies that screen the whole genome

Can “go fishing”

<table>
<thead>
<tr>
<th>Loss of DNA fragments</th>
<th>Deletion</th>
<th>Together known as COPY NUMBER VARIANTS (CNVs)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Gain of DNA fragments</td>
<td>Duplication</td>
<td></td>
</tr>
</tbody>
</table>
Microarray results

Possible test results

1) No copy number variant is found
   or the CNV is thought very unlikely to be significant (size, position, no known genes)

2) A copy number variant is found that explains the health or developmental concern.

3) A copy number variant is found but its impact on health or development is unclear.

4) A copy number variant is found that contains a gene or genes unrelated to the developmental or health concerns, but which could potentially cause other health problems in the future
<table>
<thead>
<tr>
<th>Microarray results</th>
<th>Frequency</th>
<th>Interpretation</th>
</tr>
</thead>
<tbody>
<tr>
<td>No CNV identified</td>
<td>65-70%</td>
<td>The cause of the patient’s ASD/DD/ID remains unexplained.</td>
</tr>
<tr>
<td>Non-pathogenic CNV (may not be reported)</td>
<td>5-10%</td>
<td>The CNV reflects the normal variation in DNA that exists between different individuals. The cause of the patient’s ASD/DD/ID remains unexplained.</td>
</tr>
<tr>
<td>CNV of uncertain clinical significance</td>
<td>5-7%</td>
<td>The CNV’s disease causing nature is unclear. It may or may not be the cause of the patient’s ASD/DD/ID</td>
</tr>
<tr>
<td>Pathogenic CNV</td>
<td>15-20%</td>
<td>All available evidence indicates the CNV is the cause of the patient’s ASD/DD/ID.</td>
</tr>
<tr>
<td>Unexpected</td>
<td>&lt;1%</td>
<td>The CNV is not the cause of the patient’s ASD/DD/ID, but it may cause other health problems in the future. The cause of the patient’s ASD/DD/ID remains unexplained</td>
</tr>
</tbody>
</table>
What microarrays don’t do?

• Not single gene disorders
  – e.g.
    • FRAX
    • Haemophilia
    • Duchene's Muscular dystrophy
    • Neurofibromatosis

• Not imprinting (yet)
  – e.g.
    • Angelmans/Prader willi
Early intervention

- Some variation around country
- Ministry of Education/Child Development Services plus some private providers
- Funded through MOH and MOE
- Typically
  - SLT and/or EIT
  - Education support worker
  - May have VDT or OT or PT
Autism Spectrum Disorder
From dyad to triad and back

Kanner described two essential features of autism:

• a lack of interest in the social world
• a group of behaviors he referred to as ‘resistance to change’
  or ‘insistence on sameness’

Autism in DSM-5: progress and challenges
Fred R Volkmar* and Brian Reichow
http://www.molecularautism.com/content/4/1/13
Triad of Impairments

Social Relationships

Social Communication

ASD

Rigidity of Thought, Behaviour and Play
(Social Understanding)
Pervasive Developmental Disorders
(DSM-IV, 1994)

- Autistic Disorder
- PDD-N.O.S.
- Rett’s Disorder
- Asperger’s Disorder
- Childhood Disintegrative Disorder
ASD in DSM 5

DSM-5 and autism spectrum disorders (ASDs): an opportunity for identifying ASD subtypes

Rebecca Grzadzinski, Marisela Huerta, Catherine Lord
Molecular Autism 2013, 4:12 (15 May 2013)
DSM 5
Social Communication

A: Persistent deficits in social communication and social interaction across multiple contexts, not accounted for by general developmental delays, as manifest by the following, currently or by history (examples are illustrative; not exhaustive):

1. Deficits in Social-Emotional Reciprocity - RANGING
   – Abnormal social approach & failure of normal back & forth conversation
   – Reduced sharing of interests, emotions or affect
   – Failure to initiate or respond to social interaction
DSM 5
Social Communication - continued

(and )

2. Deficits in nonverbal communicative behaviours used for social interaction- RANGING
   – Poorly integrated verbal and non-verbal communication
   – Abnormalities in eye contact and body language or deficits in understanding and use of non-verbal communication
   – Total lack of facial expression and non verbal communication
DSM 5

Social Communication - continued

(and )

3. Deficits in developing, maintaining and understanding relationships, appropriate to developmental level (beyond caregivers) - RANGING

- Difficulties adjusting behaviour to suit different social contexts
- Difficulties in sharing imaginative play
- Difficulties in making friends
- Absence of interest in people
**DSM 5**

**Restricted, repetitive patterns of behaviour, interests**

**B:** Restricted, repetitive patterns of behaviour, interests, or activities as manifested by at least 2 of the following, *currently or by history* (examples are illustrative; not exhaustive):

- **Stereotyped or repetitive motor movements, use of objects or speech,**
  - *(e.g., simple motor stereotypies, lining up toys or flipping objects, echolalia, idiosyncratic phrases)*

- **Insistence on sameness, inflexible adherence to routines, ritualised patterns of verbal or nonverbal behaviour**
  - *(e.g., extreme distress at small changes, difficulties with transitions, rigid thinking patterns, greeting rituals, need to take same route or eat same food very day previously repetitive questioning).*
DSM 5
Restricted, repetitive patterns of behaviour, interests

- Highly restricted, fixated interests that are abnormal in intensity or focus
  - (such as strong attachment to or preoccupation with unusual objects, excessively circumscribed or perseverative interests).

- Hyper or Hypo reactivity to sensory input or unusual interest in sensory aspects of environment;
  - (such as apparent indifference to pain/heat/cold, adverse response to specific sounds or textures, excessive smelling or touching of objects, fascination with lights or spinning objects).
C. Symptoms must be present in early developmental period (but may not become manifest until social demands exceed limited capacities or may be masked by learned strategies in later life).

D. Symptoms cause clinically significant impairment in social, occupational or other important areas of current functioning.

E. Not better explained by Intellectual disability or global developmental delay.

   - Intellectual disability and autism spectrum disorder frequently co-occur;
   - to make comorbid diagnoses of autism spectrum disorder and intellectual disability; social communication should be less than that expected for the general developmental level.
## Severity Ratings

<table>
<thead>
<tr>
<th>ASD Severity level</th>
<th>Social communication</th>
<th>Restricted interests &amp; repetitive behaviours</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Level 3</strong> ‘Requiring very substantial support’</td>
<td>Severe deficits in verbal and nonverbal social communication skills cause severe impairments in functioning; very limited initiation of social interactions and minimal response to social overtures from others.</td>
<td>Preoccupations, fixated rituals and/or repetitive behaviours markedly interfere with functioning in all spheres. Marked distress when rituals or routines are interrupted; very difficult to redirect from fixated interest or returns to it quickly.</td>
</tr>
<tr>
<td><strong>Level 2</strong> ‘Requiring substantial support’</td>
<td>Marked deficits in verbal and nonverbal social communication skills; social impairments apparent even with supports in place; limited initiation of social interactions and reduced or abnormal response to social overtures from others.</td>
<td>RRBs and/or preoccupations or fixated interests appear frequently enough to be obvious to the casual observer and interfere with functioning in a variety of contexts. Distress or frustration is apparent when RRB’s are interrupted; difficult to redirect from fixated interest.</td>
</tr>
<tr>
<td><strong>Level 1</strong> ‘Requiring support’</td>
<td>Without supports in place, deficits in social communication cause noticeable impairments. Has difficulty initiating social interactions and demonstrates clear examples of atypical or unsuccessful responses to social overtures of others. May appear to have decreased interest in social interactions.</td>
<td>Rituals and repetitive behaviours (RRB’s) cause significant interference with functioning in one or more contexts. Resists attempts by others to interrupt RRB’s or to be redirected from fixated interest.</td>
</tr>
</tbody>
</table>
Recognition and referring
Initial Identification

• Parents often know something is different by 18 months.
• Parents are more likely to raise concerns than other health and education professionals.
• Early detection and intervention - best outcomes.
• Valid clinical diagnosis possible by 2 - 3 years
Absolute Indicators for Immediate Assessment in preschoolers:

- No babbling or pointing or other gesture by 12 months
- No single words by 16 months
- No 2-word spontaneous non-echolalic phases by 24 months
- Any loss of ANY language or social skills at ANY age
Recognition and referring
Assessment Referral pathways

• *Discuss with local DHB ASD Coordinator*
• 2-3 main routes in 0-8 year olds:
  – Preschool refers to MOE
  – GP refers to Paediatrics
  – GP/Other professional refers to Child development service
• Older primary and/or adolescents
  – CAMHS
  – May also be through paediatrics to 15 years in some areas
Components of ASD Assessment

- Developmental and family history
- Observations across more than one setting
- Cognitive assessment
- Communication assessment
- Mental health
- Behaviour
- Needs and strengths of all family members
- Physical examination
Bare minimum?

• Good history
• Information from across settings and over time
• Consistent with observations of child
• Meets DSM 5 criteria
• Not better explained by another diagnosis*
• “Best-fit diagnosis for now” and willingness to review
Differential Diagnosis in ASD

• Normal population

• Intellectual Disability

• Other neurodevelopmental disorders
  – ADHD,
  – Dyspraxia
  – Social communication disorder

• Other Psychiatric conditions
  – Anxiety, OCD, Selective mutism
When diagnosis is not clear

• Additional Assessments
  – ASD specific
    • ADI-R
    • ADOS
    • Etc
  – Cognitive assessment
  – SLT Assessment
  – Child psychiatry review
  – OT assessment
Changing Pattern of Diagnosis

• Historically autism thought to be a rare condition (2–4/10,000)

• In 1990s and early 2000s, studies reported rise in incidence of ASD in preschoolers (prevalence 60/10,000)

• Currently, the wider spectrum of ASD is thought to affect about 1% of the population (or more than 40,000 New Zealanders)
Why the increase?

- Changes in diagnostic criteria over time
- Differences in methods used in studies
- Increasing awareness amongst professionals and the wider community

- Recognition that ASD:
  - occurs in association with other conditions (e.g., ID, physical disability, syndromes, psychiatric conditions)
  - can occur in people with high IQ
  - presentation can be subtle.

- The question as to whether there has been a genuine increase remains open
Post diagnosis

- DHB ASD Coordinator
- Early intervention (preschool)
- School based support?
  - RTLB, Possibly SLT, Tips for Autism
- Parent Education
  - ASD plus
  - Tips for Autism
  - Growing up with Autism
- CDA
- Behaviour Support services – Explore or IDEA ASD services
- Advocacy organisations
  - (Autism NZ, Altogether Autism)
- OT referral through Child development service
Post diagnosis –
Social Stories

Books about George

I can go to the hairdresser to get my haircut

George gets a haircut
Other visual aids
Medications in Autism

• (None)

• Can be useful for **associated symptoms**
  1. Anxiety
  2. Inattention and impulsivity
     i.e Co morbid diagnosis ADHD
  3. Self harm or serious aggression refractory to behavioural intervention
  4. Sleep
Melatonin

- Recently provisionally approved by Pharmac for sleep disorders in ASD/ID etc
- No date for roll out
- Will probably only be Circadin which is not controlled release when crushed
- Currently section 29
- Dose
  - 1.5 to 3mg standard release
  - 2-4 mg controlled release
- $$$$ -
  - $2-3 per day
- Useful when effective
- Trial off after 2-6 weeks
  - and again changing back from Daylight saving if unsuccessful
Prognosis

• Dependent on IQ and language level attained
• Difficult to predict at 2-3 years

• As a general rule
  – The majority show some improvement over time
  – Community participation and independence remains less than predicted for level of academic attainment
A Communication trajectories

- High (12.7%)
- Bloomers (7.5%)
- Medium-high (25.8%)
- Medium (24.5%)
- Low-medium (18.4%)
- Low (11.1%)

Axes:
- Y-axis: High to Low
- X-axis: Age (3 to 14)

Graph shows the progression of communication functioning across different age groups.
C Repetitive behavior trajectories

- Never (21.4%)
- Improving (8.1%)
- Declining (7.1%)
- When stressed (28.0%)
- Daily (27.6%)
- Usually (7.8%)

Functioning

Age

High

Low
KEY SIGNS OF ASD IN CHILDREN AGED 4 – 8 YEARS
Communication impairments

• Abnormalities in language development, including muteness, odd or inappropriate intonation patterns, persistent echolalia, reference to self as ‘you’ or ‘she/he’ beyond 3 years, unusual vocabulary for child’s age/social group

• Limited use of language for communication and/or tendency to talk freely only about specific topics
Social impairments

- inability to join in with the play of other children, or inappropriate attempts at joint play (may manifest as aggressive or disruptive behaviour)

- easily overwhelmed by social and other stimulation

- failure to relate normally to adults (too intense/no relationship)

- showing extreme reactions to invasion of personal space and extreme resistance to being ‘hurried’. 
Restricted interests and repetitive behaviours

- lack of flexible, cooperative imaginative play/creativity (although certain imaginary scenarios, e.g., copied from videos or cartoons, may be frequently re-enacted alone)

- difficulty in organising self in relation to unstructured space (e.g., hugging the perimeter of play area)

- inability to cope with change or unstructured situations, even ones that other children enjoy (e.g. kindy trips, teachers being away)

- preoccupation with restricted patterns of interest that are abnormal either in intensity or focus; over-attention to parts of objects.
Restricted interests and repetitive behaviours

- Over- or under-sensitivity:
  - **sound** (e.g., has trouble functioning when there is noise around), touch (e.g., difficulties standing in line or close to others, avoids getting messy, or excessively touches people and objects)
  - **movement** (e.g., avoids playground equipment or moving toys, or seeks all kind of movement, and this interferes with daily routines)
    - **visual stimuli** (e.g., prefers to be in the dark, discomfort or avoids bright lights)
    - **smells** (e.g., deliberately smells objects)

- Unusual responses to movement (e.g., toe walking and hand flapping)
Other features sometimes seen
- not explicitly mentioned in DSM 5

- unusual profile of skills/deficits (e.g., social and motor skills very poorly developed, whilst general knowledge, reading or vocabulary skills are well above chronological/mental age)

- any significant history of loss of skills.
Adolescent Presentations

Similar presentations to younger children plus:

• Growing awareness of social deficits

• Abstract thought issues

• Organisational difficulties

• Depression, social isolation (+/- bullying)
Adolescent Presentations

- Suicidal ideation
- Identity issues
- Sexual issues and fixations
- Unusual forensic presentations
- Impairment associated with symptoms in multiple domains
Older Children and Teens

• Diagnosis triggered by:
  – symptom changes and diagnostic dilemmas (what once made sense doesn’t any more)
  – social deficits becoming more obvious
  – difficulty meeting academic expectations