Diagnosis of Autism: Past, Present and Future

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Outline

• Historical perspective
• Core deficits
• Problems of definition
• Future perspectives
• Evidence versus pragmatism
Occasionally Her "I don't know what to wear today" clothing crisis would get completely out of hand.

**PROBLEM 1:**
Life is full of uncertainties
PROBLEM 2: The professionals don’t know everything!
PROBLEM 3: Information overload, variable quality
Journey to Present Framework
MAKING SENSE OF THE AUTISTIC SPECTRUM

Kanner

Creak

Kolvin

Asperger

CORE PROBLEM

Wing & Gould (1979)

CORE IMPAIRMENT IN
SOCIAL INTERACTION

TRIAD OF IMPAIRMENTS
DIAGNOSIS ON ICD-10

- Social interaction deficits
- Social communication deficits
- Rigid behaviours or thinking / obsessions
WHAT MAKES DOCTORS HAPPY?

Hypothyroidism

Hormone deficit known

Screening programme

Early Diagnosis

Institute Treatment

Clear biological basis

Proactive approach

Biological test

Abort catastrophic effect
PROBLEMS IN AUTISM

• No biological markers
• Genetics still poorly understood
• Screening problematic
• Emerging clinical picture over time
• No miracle cures
DEFINING SYNDROMES

Syndrome: Random collection of quirks and oddities that, if given a name, will be readily diagnosed by doctors who will then think they’ve achieved something.

A Sceptic’s Medical Dictionary
Michael O’Donnell
Problems of Syndrome Definition:

Children don’t often fit into neat boxes!

Nature never draws a line without smudging it (Lorna Wing)
AUTISM AS A DIMENSIONAL DISORDER

• Categorical definitions have limited and specific uses
  ⇒ Making clinical diagnoses
  ⇒ Supporting research

• Biologically – autism is a dimensional disorder – a neurobehavioural endpoint of many pathologies
Problems in social interaction
MANIFESTATIONS OF SOCIAL IMPAIRMENT

• Children with autism can:
  ⇒ Show attachment behaviours
  ⇒ Be socially approaching
  ⇒ Give eye contact

• Children with autism may have particular problems in:
  ⇒ Sharing and directing attention
  ⇒ Recognising affect
  ⇒ Understanding subtleties of social situations

Decreasing severity
“What worries me is that when Jonathan leaves school he’ll have 6 or 7 GCSE’s, but he won’t be able to go out and buy himself a shirt.” (Age 15)

“David knows that when he walks into the tennis club he ‘gets it wrong’, whilst his younger brother Peter is fine. He doesn’t understand why this is and he gets very depressed.” (Age 11)
PRESENTATION OF HIGH FUNCTIONING GROUP

- Often (but not always) present after age 3
- With hindsight, ‘peculiarities’ present from under age 3
- Often behavioural rather than language-led presentation. For example:
  - ‘Hyperactive’
  - Social problems in school
  - School exclusion
  - School ‘failure’
Problems in social communication
LANGUAGE PRECURSORS

• 4 - 7 months
  ⇒ mutual attention
  ⇒ pseudo conversations

• 8 - 14 months
  ⇒ social referencing (triadic vision)
  ⇒ pointing for interest
  ⇒ attachment behaviours
  ⇒ modifies behaviour in relation to other people’s emotional behaviour (e.g. smiles / frowns)
WHAT FOLLOWS FROM JOINT ATTENTION AND JOINT ACTION?

• Referential understanding is pivotal factor in vocabulary growth and pragmatic language use

• Behaviour of caregiver influences development
   ⇒ Attending to communication acts increases them
   ⇒ Expansion from child’s base (‘semantic contingency’) leads to more vocabulary
DEVELOPMENT OF LANGUAGE

• Dependent on symbolic representation

• Generative language - involves understanding of words, novel use
  ⇒ The categorisation of the world through words (over-generalisation initially - all animals ‘dog’ etc.)
  ⇒ Learning grammar creatively
Over-generalisation is understandable
Unfortunately, animals sometimes lack the necessary skills to communicate with each other.
And then there are the obsessional and rigid behaviours
Some of mother’s habits were frankly obsessive: checking the Persian rug for mites was just one of her daily routines.
Deriving categories in dimensional disorders
Where do you draw the line?
TO LUMP OR TO SPLIT?

Atypical autism
Kanner
Asperger’s
Low-moderate autism
HFA

Disintegrative disorder
Autistic spectrum
PDDs

Childhood autism
Atypical autism
Asperger’s

[From Tsai]
Distribution of Social Responsiveness Scale (SRS) scores as a function of sex (n = 1576).
Understanding autistic brains
The SCQ in an unselected (mainstream) population
N=411 (44% return)

ASD cut-off >=15; autism cut-off >=22
Additional Diagnostic Challenges
DISORDERS WITH POSSIBLE FRONTAL PATHOLOGY

• Autism
• Aspergers
• ADHD
• Dyspraxia
• DAMP
• Tourette’s etc.

Common to all - social cognition deficits
COMORBIDITY, NOT AN ‘EITHER / OR’….

• Disorders common in AS / autism
  ⇒ ADHD (prepubertal children)
  ⇒ Depression (adolescents and adults)
  ⇒ Affective disorder
  ⇒ Anxiety
SYNDROMES OF ‘DISINHIBITION’

ADHD

Frontal lobe (impulsivity)

- Distractibility
- Fidgetiness
- Unfocused attention
- Poor activity control

Tourette’s

Frontal lobe + basal ganglia (movement disorder)

OCD

DISINHIBITION

- Obsessive thoughts
- Poor task focus
- Less distractibility
- Fast associations
OTHER DISINHIBITION SYNDROMES

The David returns from a trip to the USA
Importance of impulse inhibition
Functionally impairing social deficit currently

- No
  - Functionally impairing social deficit previously
    - No
      - In wrong clinic
      - Meets criteria in 3 domains
        - Autism
    - Yes
      - Watchful waiting
      - Meets criteria < 3 domains
      - Autistic spectrum
  - Yes
    - Consider differential diagnoses of social impairment
      - No
        - Clinical assessment +/- ADI and ADOS
          - Meets criteria < 3 domains
          - Autistic spectrum
        - Other diagnosis
      - Yes
        - Sub-clinical scores on formal tests
          - ?
          - Other diagnosis
IS DSM-V GOING TO HELP US?

Severity rating + more simplified diagnostic framework
PROPOSALS (1): Diagnosis

• One spectrum called Autistic Spectrum Disorder
• No differentiation of PDD-NOS, Asperger syndrome etc.
• Diagnosis purely on behavioural symptoms
• Can occur with any other diagnosis
  ⇒ Rett syndrome
  ⇒ Intellectual disability
  ⇒ Language disorder
PROPOSALS (2): Two domains instead of three

- **Social communication**
  - Basic deficits in verbal & non-verbal communication
  - Lack of social reciprocity
  - Lack of interest or difficulty establishing relationships with peers

- **Fixated interests, repetitive behaviours**
  - Repetitive behaviours, sensory anomalies, mannerisms etc.
  - Insistence on sameness, rituals
  - Fixated interests
PROPOSALS (3): Exemplars for different age / language levels

• Social communication
  ⇒ Preschool / Child / Adolescent
  ⇒ Non-verbal or single words / phrases / fluent

• Fixated interests / repetitive behaviours
  ⇒ Preschool / Child / Adolescent
  ⇒ Non-verbal or single words / phrases / fluent
<table>
<thead>
<tr>
<th>PROPOSED DSM-V SEVERITY</th>
<th>Social Communication</th>
<th>Fixated Interests and Repetitive Behaviors</th>
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</thead>
<tbody>
<tr>
<td>Most severe ASD</td>
<td>Minimal or no social communication</td>
<td>Nearly constant, complete preoccupation, strongly resists interference with ritual</td>
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<tr>
<td></td>
<td>Some social communication but interactions noticeably disturbed</td>
<td>Frequent and interfering rituals, repetitive behaviors and fixated interests</td>
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<tr>
<td>Less severe ASD</td>
<td>Clear impairments in social communication. Meets all diagnostic criteria including symptom severity greater than threshold</td>
<td>Occasional rituals, repetitive behaviors and fixated interests; some interference</td>
</tr>
<tr>
<td>Subclinical AS Symptoms</td>
<td>Has some symptoms from one or both domains but no significant interference or impairment.</td>
<td>Odd mannerisms, some excessive preoccupations but distractible, may have ritualized behaviors but they don’t interfere with daily activities</td>
</tr>
<tr>
<td>Normal Variation</td>
<td>Socially isolated or “awkward”</td>
<td>Some ritualized behaviors and preoccupations but these are normal for developmental stage and cause no interference</td>
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</table>
Evidence versus pragmatism
“Give your evidence” said the King. “And don’t be nervous or I’ll have you executed on the spot.”

This did not seem to encourage the witness at all: he kept shifting from one foot to the other, looking uneasily at the Queen, and in his confusion he bit a large piece out of his teacup instead of the bread-and-butter.

Alice’s Adventures in Wonderland, Lewis Carroll
Life is full of problems and uncertainties.
Difficult Decisions (1)

- You need a coronary artery bypass graft (CABG). There are two nearby hospitals:
  - Hospital A is a very modern, state of the art hospital and everyone locally speaks very highly of it
  - Hospital B was the original city hospital. Your uncle died having a CABG there 2 years ago, and you heard that a neighbour's brother also died there during a CABG.

- Would you
  - Have your CABG at Hospital A
  - Have your CABG at Hospital B
  - Stay in bed – it's safer
• Hospitals A and B are required to publish their CABG mortality rates. You look them up:
  ⇒ Hospital A has a 5% mortality rate (i.e. 1 in 20 people having a CABG will die during the procedure)
  ⇒ Hospital B has a 3% mortality rate (i.e. 1 in 33 people having a CABG will die during the procedure)

• Now…..would you like to change your mind and
  ⇒ Have your CABG at Hospital A
  ⇒ Have your CABG at Hospital B
  ⇒ Stay in bed – it’s safer
Difficult Decisions (3)

• Hospitals A and B operate in different kinds of patients:
  ⇒ Hospital A is a specialist centre and takes all the ‘high risk’ patients, with the worst prognosis for surgery, which accounts for its higher mortality rate
  ⇒ Hospital B’s mortality rate is about standard for people at lower risk like yourself

• Maybe time to think again? Would you
  ⇒ Have your CABG at Hospital A
  ⇒ Have your CABG at Hospital B
  ⇒ Stay in bed – it’s safer
FLUCTUATING TIME COURE

• A. Two years old – not talking, self-directed, poor joint attention, flitting between activities.

• B. Four years old – fluent, reciprocal conversations, socially responsive, turn-taking, has formed relationships in nursery, has best friend

• C. Nine years old – socially odd and gauche, obsessed with maths and equations, tolerated by other boys, mothered by girls.
MAKING PREDICTIONS AGAIN
A FLUCTUATING BASE

Severity of feature

Child’s level
Diagnostic cut-off
Functional impairment

A
B
C

Age
Some summary thoughts

• Autism is a disorder with some clear biological substrates
• Diagnosis is not based on biologically derived criteria, but on an operational consensus
• Operational definitions have both value and limitations
• Living with disorders of social cognition is a reality; it may not always be adequately described by operational definitions
• Professionals, not biology, create boundaries and constraints.
"I will now take questions from the floor"

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