Social cognitive development in children with autism spectrum disorders: Implications for everyday life

Congrès: L'autisme, jour après jour
Parc des Expositions de Bruxelles
Samedi 25th Octobre 2008
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UCL Institute of Child Health

Outline
- What is autism?
- How our understanding of autism has changed
- How common is autism?
- What are the earliest signs of autism?
- How early can autism be diagnosed?
- Developmental theory and early interventions
- Providing answers to complex questions
  - Challenges to science and to clinical practice

What is autism?
- Difficulties in social relationships
  - Failure to use eye gaze & gesture
  - Failure to develop friendships
  - Lack of social/emotional understanding
  - Lack of sharing
- Difficulties in communication
- Repetitive and rigid behaviours

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  - Preference for routines
  - Motor mannerisms
  - Sensory abnormalities
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Nature never draws a line without smudging it... (Lorna Wing)

- Classic 'Kanner' autism
- Children with some but not the full complement of symptoms
  - 'Atypical autism'/Pervasive developmental disorder (PDD)
- High functioning autism/Asperger syndrome
- Now understood to cover a 'spectrum' of severity
- Terminology used differently in different arenas
  - e.g. Education vs. Health vs. Science
  - Can lead to confusion for parents and practitioners

Our understanding of autism in the 1980s

- Autism was considered a rare condition
  - 4 - 5 children per 10,000 i.e. 1 in 2,000 children
  - ~20 per 10,000 children with the 'triad' of impairments i.e. 1 in 500 children
  - Autism was rarely diagnosed until age 3-4 years
- Outcome in most cases was poor
- Autism was a 'unitary' disorder with strong heritable genetic underpinnings

Our understanding of autism now

- Autism is a relatively common condition
  - 100 in 10,000 of children have some form of autism i.e. 1 in a 100 children
- Autism can be reliably diagnosed by the age of 2 years in some cases
- Outcome is very variable and may depend on treatment
- Now understood as a spectrum of conditions that have both heritable and sporadic genetic underpinnings + unknown environmental factors?
  - 'autism spectrum disorders' (ASDs)
  - 'the autisms'
How common is autism?

- Picture until the late 1990s
- "Classic" figure of 4 - 5 per 10,000
  - UK: Lotter (1966), Wing & Gould (1979)
  - USA: Ritvo et al. (1989)
  - Europe: Fombonne et al. (1992, 1997)
- More recent figures nearer 10 per 10,000
- Reviews and meta-analyses
  - Fombonne (1999), Gillberg & Wing (1999)
  - 'Best estimate' for autism ~10 per 10,000
  - 'Best estimate' for all ASDs ('triad of impairments') ~20 per 10,000 (Wing & Gould, 1979; Camberwell study)

3 studies published in 2000/2001

- Baird et al. (2000) - follow-up of sample screened with CHAT at age 7-8 years in South Thames
- Chakrabarti & Fombonne (2001) - 3 to 7 years best practice surveillance study in Staffordshire
- Bertrand et al. (2001) - intensive case-finding study by CDC in Brick Township, NJ, USA.
- All found rates for all ASDs ~60 per 10,000
- Though differences in other characteristics
- Overlap in age of sample and methods

<table>
<thead>
<tr>
<th></th>
<th>Baird et al.</th>
<th>Chakrabarti &amp; Fombonne</th>
<th>Bertrand et al.</th>
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</thead>
<tbody>
<tr>
<td>Population size</td>
<td>16,235</td>
<td>15,500</td>
<td>8,896</td>
</tr>
<tr>
<td>Age</td>
<td>7 yrs</td>
<td>2.5-6.5 yrs</td>
<td>3-10 yrs</td>
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<tr>
<td>% Assessed</td>
<td>46%</td>
<td>95%</td>
<td>71%</td>
</tr>
<tr>
<td>Autism/10,000</td>
<td>31</td>
<td>17</td>
<td>40</td>
</tr>
<tr>
<td>Other ASDs/10,000</td>
<td>27</td>
<td>46</td>
<td>27</td>
</tr>
<tr>
<td>All ASDs/10,000</td>
<td>58</td>
<td>63</td>
<td>67</td>
</tr>
<tr>
<td>% Boys</td>
<td>88%</td>
<td>79%</td>
<td>73%</td>
</tr>
<tr>
<td>IQ &lt;70</td>
<td>22%</td>
<td>26%</td>
<td>49%</td>
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3 studies published in 2005/2006

- Prevalence of ASD in 1996 birth cohort = 117 per 10,000
- Prevalence of ASD in 5-16 year olds = 90 per 10,000
Birth cohort of 57,000 children in South Thames

Prevalence of ASD in 9-10 year olds = 116 per 10,000

However, case definition affects prevalence estimates

- Narrow autism 25 per 10,000
- Childhood autism 39 per 10,000
- Other ASDs 77 per 10,000
- Total ASD 116 per 10,000

Also, case ascertainment affects prevalence estimates

- Narrow autism 25 per 10,000
- Childhood autism 39 per 10,000
- Other ASDs 77 per 10,000
- Total ASD 116 per 10,000
- Locally diagnosed 45 per 10,000

Factors that affect local identification

- 64% narrow autism; 58% childhood autism;
  23% other ASD cases had a local diagnosis
- Factors that increase prior identification:
  - Parental education
  - IQ >70 – most unidentified cases had other
    neurodevelopmental diagnosis: ID/MR, learning
disability, DLD, motor or specific learning problem
- Factors not associated with prior identification:
  - SES or income
  - Child sex
  - District

Factors that affect local identification

Summary

Prevalence of disorders of the autism spectrum in a population cohort of children in South Thames: the Special Needs and Autism Project (SNAP)

Factors that affect local identification
Has autism become more common?
- No definitive evidence that it has
- What might explain the apparent rise?
  - Wider recognition
  - Broader diagnostic concept
  - More thorough studies
  - Inclusion of children with average as well as below-average IQ in studies
  - Use of the diagnosis in previously excluded groups
  - Whilst these factors may account for most or all of the apparent increase – other explanations cannot be ruled out, including a true rise in incidence

Case definition and aetiology
- The ASD phenotype may be arrived at by a number of pathogenic routes
  - That presumably overlap somewhere in development
- The spectrum is made up of individuals with different ultimate aetiologies
- Particular biological or genetic markers will not necessarily be present in all cases
- Case definition for the whole spectrum will continue to be reliant on the behavioural picture alone

A possible direction for future epidemiological studies?

What does this mean for Belgium?
- Births per year = 110,000 (UNICEF, 2006)
  - Childhood autism (39/10,000) = 429 children
  - All ASDs (116/10,000) = 1,276 children

Context for early detection
- Autism more common than previously recognised
- Greater public awareness of autism
- Increased recognition of early signs
- Until recently diagnosis was unacceptably late
- Increasing evidence for benefits of early intervention
  - Models of secondary impacts (Dawson, Mundy)
- Recognition of re-occurrence in families and implications for genetic counselling
  - 5% of autism, up to 15% of broader disability

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Volume 39(6), June 2000, pp 694-702
A Screening Instrument for Autism at 18 Months of Age: A 6-Year Follow-up Study
CHAT screening study

- CH CHECKlist for Autism in Toddlers
- Screened 16,000 children at 18-month-olds in 10 Districts from South Thames
- Focused on ‘joint attention’ behaviours known to be impaired in young children with autism
- Health Visitor and GP screen at health check
  - Parent report: Does your child ever use his/her index finger to point, to SHOW YOU something?
  - HV/GP observation: Get child’s attention, point to an object and say “Oh look! There’s an X!” Does the child look back to see what you are pointing at?

What is joint attention?

- **Responding to an adult**
  - Following an adult’s gaze shift (‘gaze monitoring’)
  - Following an adult’s point
- **Initiating a communicative exchange**
  - Looking to an object and then back at the adult (‘checking out’)
  - Pointing to show an object to an adult
- Critical ‘precursor’ to language
- We are all experts at non-verbal communication (without knowing it)
  - Unfortunately, this is not so for children with autism
Screening for autism. How did we do?

- Showed for the first time that it was possible to prospectively identify cases of autism
- 18-month-olds who by parent report and HV observation do not show joint attention or pretend play are at high risk of autism
  - “High specificity”
- However, we detected only 19/50 (38%) cases with autism
  - i.e. we missed more cases than we identified
  - “Low sensitivity”
- Cannot recommend as a population screen but can be used as part of clinical surveillance
Screening for Autistic Spectrum Disorder in Children Aged 14–18 Months. II: Population Screening with the Early Screening of Autistic Traits Questionnaire (ESAT). Design and General Findings

Claudine Boe - Noppe Veulkers - Gunna van Doorn - Roeman van Engeland - Jim K. De Bold

Summary of ESAT instrument properties

- Higher refusal rate at both stage 1 and stage 2 cf. CHAT study
  - Are parents reluctant to consider a problem in children so young?
- Of those who failed screen II and came to assessment 25% had PDD but 100% had a developmental or psychiatric disorder
- Follow-up will test initial diagnosis and sensitivity (i.e. what proportion of cases were identified prospectively)
- Aim to re-screen whole population at 6 years

'Red flags' for autism

- No big smiles or other warm, joyful expressions by 6 months or thereafter
- No back-and-forth sharing of sounds, smiles, or facial expressions by 9 months or thereafter
- No babbling by 12 months
- No back-and-forth gestures, such as pointing, showing, reaching, or waving by 12 months
- No words by 16 months
- No two-word meaningful phrases (without imitating or repeating) by 24 months
- Any loss of speech/babbling or social skills at any age

Good web-based information for parents and professionals

http://www.firstsigns.org/

http://www.autismspeaks.org/video/glossary.php
Clinical assessments of early signs

Recent progress in earlier identification brings challenges

- Reliability of diagnosis in 2 year olds?
- Establishment of the utility and limitations of standard assessment instruments with toddlers
- Ability to indicate prognosis at such an early age?
- Diagnosis needs to be explained in such a way that parents will recognise their child
- Parents may be unable to fit the classical picture of autism to their toddler
- Clinical concept of ‘working diagnosis’
- Clinicians and parents dealing with uncertainty

CHAT screen by community nurses to identify cases

Clinic assessment and diagnosis of autism (not broader PDD) at 24 months

- 26 children seen at 3 timepoints
  - T1 aged 2 years
  - T2 aged 3 years
  - T3 aged 7 years

Youngest sample studied prospectively

Stability of diagnosis from age 2 years to age 7 years

Cross-tabulation of Initial Diagnostic Measures and Best-Estimate Diagnoses at Ages 2 and 9 Years

Outcome at 7 years of children diagnosed with autism at age 2: predictive validity of assessments conducted at 2 and 3 years of age and pattern of symptom change over time

Lord, C. et al. Arch Gen Psychiatry 2006;63:694-701

Autism symptom severity scores

- Social symptoms
- Nonverbal symptoms
- Rpt symptoms

Childhood autism
N = 26

Childhood autism
N = 22

Other ASD
N = 3

Non – ASD
N = 1

Childhood autism
N = 22

Other ASD
N = 3

Non – ASD
N = 1

Childhood autism
N = 22

Other ASD
N = 3

Non – ASD
N = 1

Childhood autism
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Other ASD
N = 3

Non – ASD
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Childhood autism
N = 22

Other ASD
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Non – ASD
N = 1
Autism symptom severity scores

Developmental trajectory of symptoms

- Different trajectory over development in different symptom domains
  - Suggests that they might be separable in terms of causes and outcomes
  - Current interest in how and why features of autism cluster together in populations and clinical samples
- Over time we see both increases (in repetitive behaviours) and decreases (in social + communication impairments) in severity
- This was for the group as a whole
  - What about for individual children...?

Outcome is very variable

- Variability increases over time
  - Some children make good progress; others make relatively little
- Increasing evidence that intervention might play a role in improving outcome
- What predicts outcome in very young children?
- What might be the appropriate targets for treatment?
Implications for services?

- Primary healthcare practitioners need to be made aware of early signs
  - Community doctors, community nurses, playgroup and kindergarten staff
- Increase demand/need for appropriate pre-school autism specific therapy, social support and educational provision
- Need to improve understanding of the appropriate behaviours to assess (and treat!) in 2- and 3-year-olds

Parent training (part focus on JA)

- Monthly sessions and daily homework tasks
- Work on task compliance and management
- Focus on joint-action, turn-taking, joint attention, imitation, use of gestures
- Daily living routines (drink, mealtimes)
- Independence skills (bathtime, dressing)
- Joint play with objects (bring and show, drawing, ball play)
- Joint play without objects (tickle, mirror games, shared action songs)
- Emphasise shared meaning

Number of words comprehended

- * p < .05 (one-tailed only)
**Communication-focused RCT**

- N=58 3-to-4-year olds (~20 per group)
- 30 minute session daily in nursery for 6 weeks
- One treatment focused on promoting JA skills
  the other on promoting symbolic play skills
- ALL children receiving 30 hours a week ABA nursery program (1:1 or 1:2)
- Language outcomes 12 months later

![Graph showing Language Outcome in Autism: Randomized Comparison of Joint Attention and Play Interventions](image1)

**Challenges of phenotypic definition**

- Presentation within individuals and within a population changes with development
- Places a heavy load on our ability to measure and establish reliable thresholds
  - Notwithstanding progress, instruments such as the ADI-R and ADOS have limitations
    - Of particular importance is the reliability with which a lower threshold can be set for the broader spectrum
- Conceptual issue: is ASD a ‘lifetime diagnosis’?
  - An individual may move back-and-forward across any diagnostic boundary over time

![Graph showing growth in expressive language](image2)

**Web-based resources for schools**

- What is autism?
- How might a child with special needs be a part of our school?
- General Strategies for Intervention
- For specific members of the School Community
- Resources
- Appendix

[http://www.autismspeaks.org/community/family_services/school_kit.php](http://www.autismspeaks.org/community/family_services/school_kit.php)
Is there a boundary between population individual differences?

Challenges of phenotypic definition

Perspective

Time to give up on a single explanation for autism

Exemplar of a complex condition

Now recognised not to be a unitary disorder
- In terms of aetiology and symptoms – ‘the autisms’
- This means that biological and genetic markers will not be present in all cases
- Case definition will continue to be reliant on the behavioural picture alone
- Presentation within individuals and within a population changes with development
- Heterogeneity is a challenge for research
  - e.g. genetics; neuroscience; search for subgroups
- Not universally see as an impairment
  - ‘Neuroautistic’ vs. ‘Neurotypical’ brain

Challenges for the future

- We need more evidence on what works (for which children?)
- Will increasing understanding of the brain, biology and genetics provide meaningful answers for children and families
- What do we know about autism in adulthood?
- How can we change policy and society for the good of people with autism?

Acknowledgements

- CHAT team: Gillian Baird, Simon Baron-Cohen, Antony Cox, Auriol Drew, John Swettenham, Sally Wheelwright
- SNAP team: Gillian Baird, Emily Simonoff, Andrew Pickles, Tom Loucas, Susie Chandler, David Meldrum
- Other collaborators: Torsten Baldeweg, Sarah-Jayne Blakemore, Patrick Bolton, Michelle De Haan, Francesca Happé, Patricia Howlin, Mayada Elsabbagh, Mark Johnson, Vicky Slonims
- Funded by: