Comorbidities: ASD & Other Developmental Disorders

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Sarah Bernard & Jeremy Turk

March 2009

Royal College of Psychiatrists Publications, London
Autism Spectrum Disorders, Intellectual Disabilities & ADHD:

- are developmental disabilities
- are not psychiatric disorders
- But they predispose individuals to mental health problems for a variety of biological, psychological, educational and social reasons
What are developmental disorders?

- Early onset
- Long-term
- Frequently multiple
- Interferences in normally fluent skill acquisitions
- Necessary for maximisation of potential and quality of life
- Producing adverse physical & psychological functional consequences
- & multiple social adversities & social disadvantage
Autistic Spectrum Disorder

A.D.H.D.

Intellectual disability
ASD Core Diagnostic Criteria:

- Impairments in reciprocal social interaction
- Impairments in language & communication
  - Receptive & expressive
  - Verbal & non-verbal
- Repetitive & stereotypic behaviours & interests
  - Gross motor
  - Abnormal sensory experiences
  - Abnormal obsessional interests
  - Insistence on routine & sameness
- Lack of imaginary & symbolic skills
Prevalence: how common?

- Kanner 1943: 3-5/10,000
- Fombonne: 10-20/10,000
- NAS: 1/200
Gender Ratio

- Autism: m:f = 2:1
- Autism Spectrum: m:f = 5:1
- Asperger syndrome: m:f = 9:1
Association Between Intellectual Disability & Autism

- 70% of children with ASD have a non verbal IQ below 70
- 50% of children with ASD have a non verbal IQ below 50
- Only 5% of children with ASD have an IQ above 100 (high functioning autism)
- Degree of intellectual disability related to likelihood of having ASD & severity of autistic features
- Up to 50% of individuals with “severe learning difficulties” have an autistic spectrum disorder
Having intellectual disability predisposes to disorders of socialisation, language, attention & concentration.

Likelihood & severity increase with increasing degrees of intellectual disability.

Having ASD or ADHD is a vulnerability factor for other specific as well as general learning difficulties.
Additional diagnostic criteria:

- Onset before 3 years of age
- Other non-specific problems
  - Phobias
  - Sleep & eating disorders
  - Tantrums & aggression
  - Psychiatric disorders
    - Depression
    - Anxiety states
    - Paranoia
    - Obsessive-compulsive disorder
    - Catatonia
Autism is Highly Genetic

- MZ/DZ twin studies
- Sibling risks
- Family prevalences (Jorde et al)
- Parental features (Piven et al)
- Broader phenotype within families
- Adoption studies
- Discrete genetic aetiologies
- N.B. Roumania
- N.B. Epilepsy
Autism may be a presenting feature of:

- fragile X syndrome (Xq27.3)
- untreated phenylketonuria (12q41.1)
- neurofibromatosis (17q11.2, 2p22)
- tuberous sclerosis (16p13.3, 12q14, 9q34)
- Williams syndrome (7q11.2)
- Angelman syndrome (Xq28, 15q11-13)
- Turner syndrome (X0)
- Cohen syndrome (8q22-23)
- Duchenne muscular dystrophy (Xp21.2)
What Sort Of Autism Is It?

- **Fragile X Syndrome**
  - Friendly & sociable (albeit shy & socially anxious) personality in presence of certain autistic-like features

- **Tuberous Sclerosis**
  - Stubborn, oppositional, defiant with frequent overactivity & inattentiveness

- **Smith-Magenis Syndrome**
  - Frequent ASD using semi-structured assessments yet rarely diagnosed as such (+ ID, ↓sleep, SIB, ADHD)

- **Angelman Syndrome**
  - High rates of predominantly aloof & passive ASD

- **Turner Syndrome**
  - Social anxiety, shyness & attentional problems
  - Ring Variant associated with severe intellectual disability & autistic features
Down Syndrome: current understanding

- Intellectual disability
- Characteristic personality & temperament
- Relatively low rates of autistic spectrum disorders & attention deficit disorders in childhood
- Depression
- Alzheimer disease
Fragile X Syndrome: Prevalence

○ Full Mutation
  - 1 per 3600 Males
  - 1 per 4000 Females

○ Premutation
  - 1 per 700 Males
  - 1 per 259 Females

SIMILAR FREQUENCIES IN ALL ETHNIC GROUPS
ONE OF THE MOST COMMON SINGLE GENE DISORDERS
THE MOST COMMON IDENTIFIABLE INHERITED CAUSE OF INTELLECTUAL DISABILITY
DNA CGG Expansions

- **Normal:** 6-40
- **Intermediate:** 40-55
- **Premutation:** 50-200
- **Full mutation:** greater than 200
Fragile X Syndrome: Intellectual functioning

- usually mild to moderate intellectual disability
- verbal/performance discrepancy
- characteristic developmental trajectory
Fragile X Syndrome: Speech & language
(Cornish, Sudhalter & Turk, 2004)

- jocular litanic phraseology
- perseveration
- repetitiveness
- echolalia
- cluttering
- sounds more rapid “but isn’t”
Fragile X Syndrome: Social impairments (Turk & Graham, 1997)

- social anxiety
- aversion to eye contact
- self-injury, usually hand biting in response to anxiety or excitement
- delayed imitative and symbolic play
- stereotyped & repetitive behaviours
Fragile X Syndrome & Autism: (Cornish, Turk & Levitas: 2007)

- 4-6% of people with autism have fragile X syndrome
- A substantial minority of people with fragile X syndrome have autism (29%)
- Many more people with fragile X syndrome have a characteristic profile of communicatory and stereotypic “autistic-like” behaviours
Distinguishing Behaviours:

- delayed echolalia
- repetitive speech
- hand flapping
- gaze aversion
- good understanding of facial expression
  (Turk & Cornish, 1998)
- Theory of mind as expected for general levels of ability (Garner, Callias & Turk 1999)
- friendly and sociable but may be shy
Boys with Fragile X Premutations: (Aziz et al., 2003)

Rates of:

- Delayed development of adaptive behaviours
- Autistic spectrum disorders
- Attention deficit disorders
- Speech & language problems
  - Social use of language
  - Speech intelligibility
- Expressive language
Men with Fragile X Premutations: (Mills et al., 2002)

- Slow, polite & precise +++
- Overfriendly & over-compliant
- Poor at showing emotions and feelings
- Social perception & empathy problems
- Problems making & keeping close friendships
- Poor visuo-spatial skills
- Difficulties concentrating & sustaining attention
- Memory problems relating to accessing memory & forgetfulness
- Physical & psychological symptoms attributable to stress
Females with Fragile X Syndrome:
(Turk & Howlin, 2003)

- ↓ intellectual functioning range
  - Mean = 60, no verbal/performance discrepancy
- ↑ Autistic Spectrum Disorder
- Irrespective of diagnostic label:
  - shyness, social anxiety, self-conscious, easily embarrassed, social isolation
  - obsessional including obsessive worrying
  - problems socialising
  - Social use of language & “semantic/pragmatic problems
Causes of developmental disorders can be

- Infective
- Toxic
- Psychosocial
- Usually genetic
- …and very often have ID, ASD & ADHD as part of their profiles
Intrauterine infections

- Toxoplasmosis
- Rubella
- Cytomegalovirus
- Herpes simplex
- H.I.V (AIDS)
- Syphilis
FETAL ALCOHOL SYNDROME
(Alcohol Related Neurodevelopmental Disorder)

- most common major toxin to which fetus is exposed
- MLD, pre & post-natal growth deficiency
- IQ in MLD/borderline range
- Fine motor & visuospatial problems, tremulousness
- Executive function, numeracy & abstraction problems
- Expressive & receptive language difficulties
- Irritability in infancy, hyperactivity in childhood
- Problems perceiving social cues
- Very unstable family environments
Cerebral Palsy

- Psychiatric disorder in 40%
- No gender predominance for boys
- Hemiplegia:
  - 25% conduct/emotional disorder
  - 10% hyperkinetic disorder
  - 3% autistic disorder
- Best predictor = low IQ
- Disorders manifest identically to those of psychosocial origin
Autistic Disorder & Epilepsy in Children: Implications for the Autism Construct

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Martin Bax
Imperial College, London

Christopher Gillberg
University of Gothenburg
Inter-group comparisons

**ASD**

- **Gender**
  - 52m, 8f (6.5:1)
    
    *Chi square 6.71, p=0.010*

- **Age (months)**
  - $x = 136.8$ (SD=29.5)
    
    *t=1.83, p=0.07*

- **IQ/DQ (verbal)**
  - $x = 36.7$ (SD=25.5)
    
    *t=-1.15, p=0.25*

**ASD + Epilepsy**

- **Gender**
  - 40m, 20f (2:1)

- **Age (months)**
  - $x = 147.1$ (SD=32.3)

- **IQ/DQ (verbal)**
  - $x = 31.4$ (SD=24.9)
Age at diagnosis of ASD

![Graph showing age of diagnosis of ASD with t = 3.59, p = 0.001]
Comparison of DISCO variables:

↑ impairment in ASD with epilepsy

- Gait problems
- Clumsiness & fine motor problems
- Continence issues
- Shrieking & screaming
- Behaviour problems in public & at home
- Lack of awareness of own identity
- Abnormal eye contact: staring
- Lack of interest in peers
- Inappropriate sociability/social interactions
Comparison of DISCO variables:

↑ impairment in ASD only group

- Abnormal fascinations with objects
- Abnormal eye contact: brief glances
Developmental Trajectories

- ↑ Social skills
- ↑ Communication skills
- Imagination skills
Familial stress & child’s sleep

- **Stress:**
  - Families with children who have epilepsy & autism reported significantly greater parent and family problems.
    - U = -2.336, p = 0.020

- **Sleep:**
  - Children with epilepsy & autism were significantly more likely to have sleep breathing disorders than children with autism only.
    - U = -2.422, p = 0.015
Conclusions

- First study to compare clinical profiles of matched samples of children with ASD and ASD+E
- Significantly more equal gender ratio in ASD+E group
- ASD+E group likely to receive a later diagnosis of ASD
- Clinical profiles: ASD+E display more impaired motor skills, delayed daily living skills, more challenging behaviour in public and more staring behaviour
- Not only *degree* of impairment but *type* of impairment is different across groups
- Social interaction style – contrary to expectations
- ASD with epilepsy represents a different syndrome from ASD alone
Clinical Types

- Aloof
- Passive
- Active but odd
- Overpedantic & pseudomature
- Relates well, but only to one person
Important Dimensions

- Degree of insight
- Drive to socialise
Psychological Impairments

- General intellectual disability (common)
- Uneven cognitive profile & V/P discrepancy
- Alexithymia
- Lack of theory of mind
- Executive function deficits
- Poor central coherence
  - ↓ drive towards meaning, piecemeal processing
  - “proof reader” rather than “big picture” people
  - → world perceived in fragmented & often incomprehensible & scary way
Mental Health Problems in Children & Young People with Autism Spectrum Disorders

- Are common
- Are frequently severe, multiple & challenging
- Often manifest in different ways from usual e.g. aggression, self-injury, chaotic disruptive & destructive hyperactivity, repetitive stereotypic behaviours, obsessions, passive resistance
- Create substantial morbidity for family
- Have substantial economic cost
- Are treatable
“We are dealing with spectra of disorders...the reasonable ways forward would be to retain umbrella diagnoses for “ASD, ID & ADHD” and to specify according to:
- Severity & nature of defining features
- General level of intellectual functioning
- Unusual or specific clinical traits
- Associated medical conditions
  - causative, consequential, complicating, coincidental
- Associated specific developmental difficulties
- Associated social issues
- Possible aetiologies
- Management intervention & support implications