

# **Autistic Spectrum Disorders: A Research and Policy Update**

**Matt Eggleston**

**2010**

MAY 6, 2002

www.time.com AOL Keyword: TIME

THE COMING JOB BOOM ■ PAKISTAN'S SPY AGENCY ■ REUNION TV

# TIME



## INSIDE THE WORLD OF AUTISM

Tammy Barrett, 11,  
of San Jose, California,  
has Asperger syndrome

More than one million Americans  
may have it, and the number  
of new cases is exploding.  
What scientists have discovered.  
What families should know.

MAY 15, 2006

www.time.com AOL Keyword: TIME

POP! GOES SCHOOL SODA ■ EXCLUSIVE: NINTENDO'S NEW GAME

# TIME

NEW INSIGHTS  
INTO THE HIDDEN  
WORLD OF

# AUTISM

BY CLAUDIA WALLIS

Nick Firth, 9,  
of Mine Hill, N.J.



# DSM-IV Criteria for Autistic Disorder

- A. A total of six (or more) items from 1), 2), and 3), with at least two from 1), and one each from 2) and 3):
- 1) qualitative impairment in social interaction:
    - a) marked impairment in the use of multiple nonverbal behaviours
    - b) failure to develop peer relationships appropriate to developmental level
    - c) lack of spontaneous seeking to share enjoyment, interests, or achievements with others
    - d) lack of social or emotional reciprocity

# Cont.

## 2) qualitative impairments in communication:

- a) delay in, or total lack of, the development of spoken language
- b) marked impairment in the ability to initiate or sustain a conversation with others
- c) Stereotyped and repetitive use of language or idiosyncratic language
- d) Lack of varied, spontaneous make-believe play or social imitative play appropriate to developmental level

3) Restricted repetitive and stereotyped patterns of behaviour, interests, and activities:

- a) encompassing preoccupation with one or more stereotyped and restricted interests, abnormal in intensity or focus
- b) inflexible adherence to specific, non-functional routines or rituals
- c) stereotyped and repetitive motor mannerisms
- d) Persistent preoccupation with parts of objects

B. Delays or abnormal functioning prior to age three years

C. Not better accounted for by Rett's Disorder or Childhood Disintegrative Disorder

# DSM-V ASD?

- Must meet criteria 1, 2, and 3:
- 1. Clinically significant, persistent deficits in social communication and interactions, as manifest by all of the following:
  - a. Marked deficits in nonverbal and verbal communication used for social interaction:
  - b. Lack of social reciprocity;
  - c. Failure to develop and maintain peer relationships appropriate to developmental level
- 2. Restricted, repetitive patterns of behavior, interests, and activities, as manifested by at least TWO of the following:
  - a. Stereotyped motor or verbal behaviors, or unusual sensory behaviors
  - b. Excessive adherence to routines and ritualized patterns of behavior
  - c. Restricted, fixated interests
- 3. Symptoms must be present in early childhood (but may not become fully manifest until social demands exceed limited capacities)

# Epidemiology

Earlier studies 1960s and 1970s	0.5/1000
Later studies (median 30 studies)	1/1000
2007 American Academy Paediatrics	6/1000
UK (Baird et al, 2006; Baron-Cohen 2009)	1/100
Male:Female	4:1
No effect of social class	

# Asperger's in girls

THE PRESS 5.5.09

Asperger's is a condition that has long been known to affect boys, who may have obsessive interests or struggle to make friends. Now an expert says many more girls have it than was thought. AMELIA HILL reports.

**D**octors in Britain are failing to diagnose thousands of girls who have Asperger's syndrome, says one of the world's leading experts.

Dr Judith Gould has accused the medical world of missing and overlooking girls with the condition, condemning them to lives of such misery that many resort to extreme self-harm and anorexia.

Gould and colleague Lorna Wing carried out ground-breaking research into the link between Asperger's syndrome, autism and other pervasive developmental disorders in 1979. Exploiting that insight, they pioneered the concept of the autism spectrum. Now Gould, a children's consultant

Boys go into attack mode when frustrated, while girls suffer in silence and become passive-aggressive. Girls learn to appease and apologise.

Tony Attwood  
*Asperger's clinician*

diagnosis in the first place can often feel like an insurmountable hurdle, with many doctors unaware that the condition can affect females.



Syndrome: Dove Charbon played by Shelley Long in the TV sitcom *Cheers* displayed Asperger's characteristics, such as performative speech and poor social skills.

# Aetiology : Genetics

Twin Studies MZ : DZ 60-90 : 5

Heritability estimate Over 90%

Family History Autism 2-3%; PDD 5%; Broader Phenotype 20%

Also MDE; Anxiety; Social Difficulties

"Idiopathic" versus "Secondary" Autism


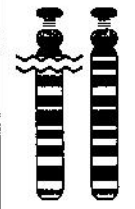
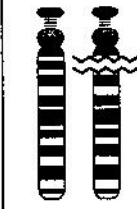
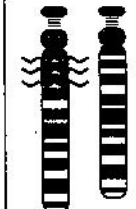

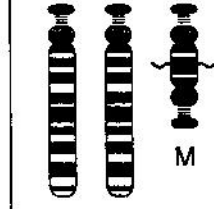
# “Secondary” Genetic Autism

- Fragile-X 3-4%
- Tuberous sclerosis (Chr 6 & 9) 2-3%
- Isodicentric (Idic) Chr 15 1%

# Idic 15

- Chromosome Region 15q 11q 13 known for its instability
- Maternal inverted duplication of 15q 11q 13 in 1% of individuals with ASD
- ASD; hypotonia; seizures; intellectual disability
- Paternally - derived deletion → Prader-Willi Syndrome
- Maternally – derived deletion → Angelman Syndrome

A.

Normal	Paternal Deletion	Maternal Del	Paternal Dup	Maternal Dup	Maternal inv dup(15)
					
P M	P M	P M	P M	P M	P M
Normal	PWS	AS	Non-autistic	Autistic Features	Autism

# Idiopathic Autism

Molecular genetic methods : all but 2 chromosomes linked

Linkage studies : 7 q (but different loci); 17 q

Cytogenetic studies : 15 q 11-13 ; long arm 2 and 7

Candidate gene studies : EN2 (Engrailed 2; cerebellar development)  
SLC 6A4 ? (serotonin transporter)

Maternal and Paternal age

Spontaneous deletions and duplications : 10%?

Gene x Gene and Gene x Environment

# Neuropathology

Macrocephaly in early  
childhood

20-30% > 2 SD

White matter and grey  
matter increase

Reduced Purkinje cells and Granule cells in  
cerebellum

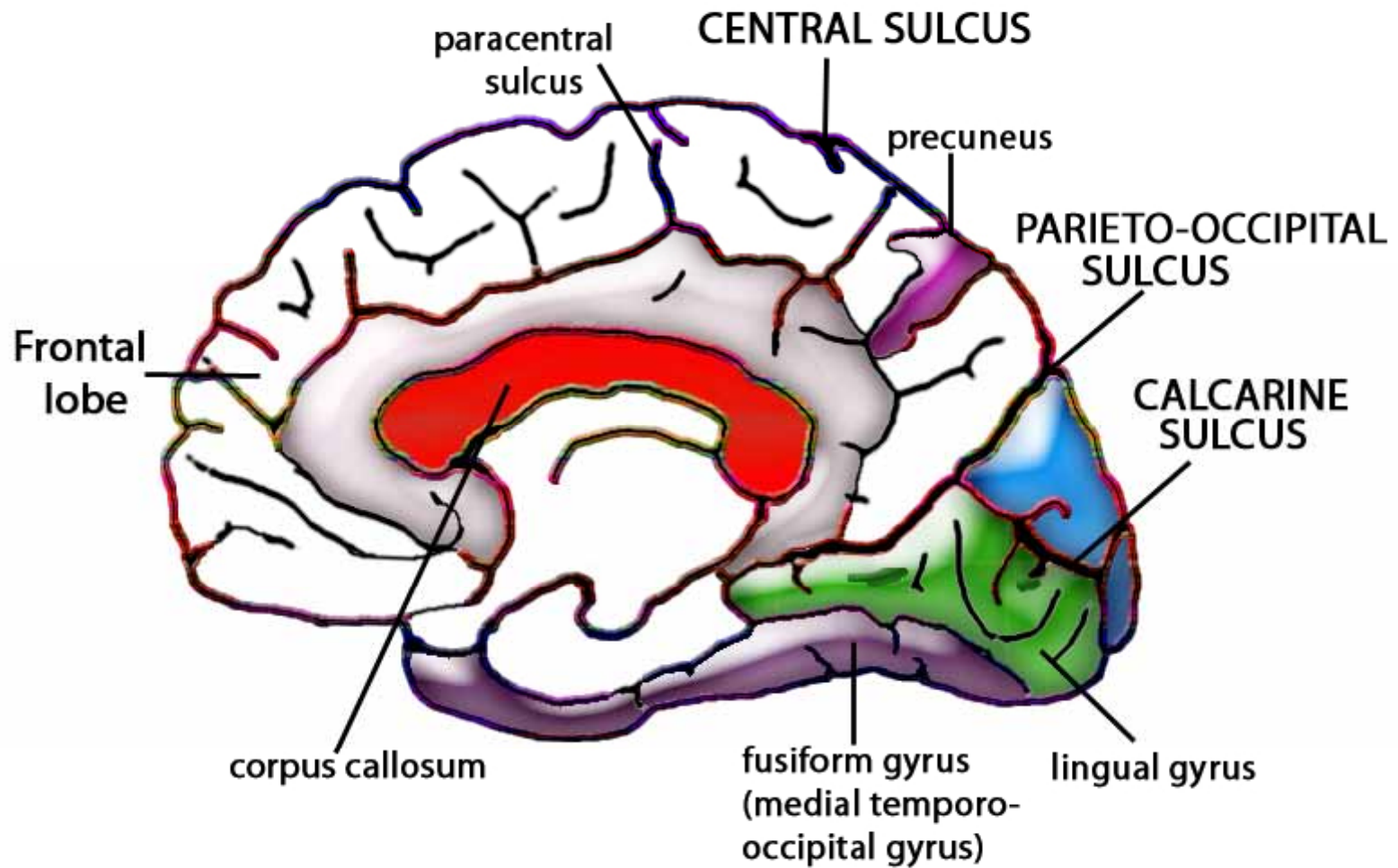
Minicolumn abnormalities in cortex

Reduced limbic neuronal size, dendritic  
arborization

# Imaging

- Underactivation fusiform gyrus on face perception tasks
- Amygdala hypoactive on social and affective judgement tasks
- Reduced mirror-neuron system (pars articularis) activation on observation or imitation of facial expressions
- Lower functional connectivity among cortical regions on complex tasks

Diagrammatic mid-sagittal view of the brain showing the right hemisphere



# Treatment

- Promote best functioning taking into account strengths and weaknesses based on comprehensive assessment.
- Early, highly structured individualised intervention plan
- Education intensive and highly structured, predictable.
- Visual planning
- Overt social curriculum
- Speech and language focus on communication
- Occupational therapy
- Behaviour therapy
- No clear evidence any particular model favoured over another. (Applied Behavioural Analysis versus Developmental models.)

# Pharmacotherapy Repetitive Behaviour, Rigidity, OC

- Clomipramine (Gordon et al, 1993)
- Fluoxetine (Hollander et al, 2005)
- Not citalopram! (King et al, 2009)
- Risperidone (McDougle et al, 2005)
- Valproate (Hollander et al, 2006)

# Pharmacotherapy : ADHD Symptoms

Stimulants

RUPP, 2005\*

Quintana et al, 1995

Harden et al, 2000

Clonidine

Fankhauser et al, 1992

Jaselskis et al, 1992

# Pharmacotherapy : Aggression, Irritability

- Risperidone RUPP, 2002\*  
Shea et al, 2005  
Aman et al, 2005  
Troust et al, 2005 (p w/d)  
Miral et al, 2008(>hpl)

Older antipsychotics eg Campbell 1978

# Pharmacotherapy : Sleep

- Melatonin
- Garstang and Wallis (2006)
- Wardell et al (2008)  
(neurodevelopmental difficulties)

# NZ ASD Guideline

- Casey Albury-Thompson → 1998 Curry Report
- 2002-2008 Development of the ASD Guideline  
Jointly funded by MoE and MoH
- “Aspirational” rather than realistic
- 8 Parts; 305 pages
- 3 Workstreams
- 63 Key Recommendations, 188  
Recommendations and 55 Good Practice Points

# NZ ASD Guideline

- Early identification
- Referral Pathways and Service Coordination
- Multiagency and multidisciplinary assessment (or a highly trained health practitioner)
- Comprehensive assessment including audiology
- 0-7 years developmental paediatrician/paediatrician
- Family support (flexible and timely)
- Education for 15-25 hours per week
- Vocational Planning/High standard vocational services
- Structural and predictable respite
- Training in ASD for all professionals who come into contact with ASD
- National Plan for professional learning and development
- Cultural aspects

# Some inconsistencies

Part 1 page 43:

1.2.7 The assessment of intellectual, adaptive and cognitive skills...(in) ASD should be seriously considered and, where possible and appropriate, formally assessed. (Grade B)

Appendix 6 page 293

“Formal baseline cognitive and/or developmental assessment is recommended at diagnosis”

but

3.2:d page 113

IQ tests should be used with caution

- IQ tests, when used should be accompanied by ... assessment of language ... assessment of adaptive function ...
- Cognitive assessments should be administered by a psychologist with experience and training in ASD.” (Grade A)

# Post ASD Guideline release

- Impact Analysis (not released)
- Implementation funding \$18m over 3 years (\$25 person/year or \$200,000 per DHB/year)

# NZ Guideline Group and Implementation Advisory Group

- NZGG contracted to support the implementation process
- NZGG established the IAG to advise NZGG on how to implement key recommendations of the Guideline including allocation of \$18m funding

# Priority Areas for New Funding

- Assessment and Diagnosis
- Support to Strengthen Families
- Intervention
- Respite

# Assessment and Diagnosis

- Website for “recognisers” and “referrers”
- ASD Training Workshop(s) for Specialists (Tanya Breen/Werry C)
- Autism NZ contract for resources for home and community support providers
- NASC ASD Service enhancement programmes
- Developmental Services Coordination expressions of interest (~ \$0.8m)

# Support to Strengthen Families

- SPELL evaluation
- Bright Sparks
- Tips for Autism (MoE 4/7 support plans for kids with ASD)
- ASD specific parent education (e.g. earlybird)

# Intervention

- Feasibility study for ASD specific behaviour support services
- Family and Whanau community outreach services (Auckland)
- ASD specific communication and behaviour services (Auckland; piloted 2005/6)

# Respite

- ASD specific respite models report
- \$1m over 3 years for out of home family respite services (N.I.: Spectrum care trust and Open Home Foundation)

# Living Guideline Group

- MoE
- Keep the ASD Guideline current
- Changes evidence-based
- Applied Behavioural Analysis evaluated

# Points for Discussion?

- MoH roles and responsibilities  
teleconference
- Does disability “own” ASD
- What are CAMHS roles/reponsibilities
- ASD: difference, disability or disorder?  
Implications of this for resourcing
- 1% prevalence
- Adult services??