

Assessment and diagnosis of Autism



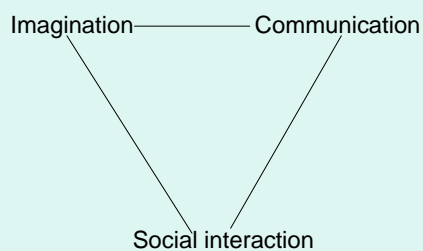
Andrew Lloyd Evans MD

Pervasive developmental disorders

- Communication
- Socialisation
- Restriction of interest



The autistic triad (Lorna Wing)



PREVALENCE OF AUTISTIC SPECTRUM DISORDERS (Baird et al. 2006)

- | | |
|--------------------|------------|
| • 'Narrow' autism | 25/10,000 |
| • Childhood autism | 39/10,000 |
| • Other ASDs | 77/10,000 |
| • Total ASD | 116/10,000 |

- Therefore 1:100 has some form of autism
- 55% IQ <70 - not in higher functioning
- 60% have 1 or more co-morbidities
- Broader autism phenotype

BUT?

- Are we over diagnosing?
- Medicalisation of educational issues
- Inability to tolerate difference
- Does educational management need a medical diagnosis?
- Descriptions not diagnoses?
- Beware collusion with parents

Why do it?

- Probably benefits severe children
- Probably benefits moderate children
- What about high functioning children?
 - Child themselves
 - Parents
 - School

You can use the classification systems:

- ICD 10 -The pervasive developmental disorders
- DSM-IV – Autistic disorder

ICD-10 Criteria for "Childhood Autism"

- A. Abnormal or impaired development is evident before the age of 3 years in at least one of the following areas:**
1. receptive or expressive language as used in social communication;
 2. the development of selective social attachments or of reciprocal social interaction;
 3. functional or symbolic play.
- B. A total of at least six symptoms from (1), (2) and (3) must be present, with at least two from (1) and at least one from each of (2) and (3)**
1. Qualitative impairment in social interaction are manifest in at least two of the following areas:
 - a. failure adequately to use eye-to-eye gaze, facial expression, body postures, and gestures to regulate social interaction;
 - b. failure to develop (in a manner appropriate to mental age, and despite ample opportunities) peer relationships that involve a mutual sharing of interests, activities and emotions;
 - c. lack of socio-emotional reciprocity as shown by an impaired or deviant response to other people's emotions; or lack of modulation of behavior according to social context; or a weak integration of social, emotional, and communicative behaviors;
 - d. lack of spontaneous seeking to share enjoyment, interests, or achievements with other people (e.g. a lack of showing, bringing, or pointing out to other people objects of interest to the individual).
 2. Qualitative abnormalities in communication as manifest in at least one of the following areas:
 - a. delay in or total lack of, development of spoken language that is not accompanied by an attempt to compensate through the use of gestures or mime as an alternative mode of communication (often preceded by a lack of communicative babbling);
 - b. relative failure to initiate or sustain conversational interchange (at whatever level of language skill is present), in which there is reciprocal responsiveness to the communications of the other person;
 - c. stereotyped and repetitive use of language or idiosyncratic use of words or phrases;
 - d. lack of varied spontaneous make-believe play or (when young) social imitative play

Diagnostic Criteria for Autistic Disorder DSM-IV

[The following is from *Diagnostic and Statistical Manual of Mental Disorders: DSM IV 299.0*
 (1)A total of six (or more) items from (A), (B), and (C), with at least two from (A), and one each from (B) and (C)
 (A) qualitative impairment in social interaction, as manifested by at least two of the following:

1. marked impairments in the use of multiple nonverbal behaviours such as eye-to-eye gaze, facial expression, body posture, and gestures to regulate social interaction	
2. failure to develop peer relationships appropriate to developmental level	
3. a lack of spontaneous seeking to share enjoyment, interests, or achievements with other people, (e.g., by a lack of showing, bringing, or pointing out objects of interest to other people)	
4. lack of social or emotional reciprocity (note: in the description, it gives the following as examples: not actively participating in simple social play or games, preferring solitary activities, or involving others in activities only as tools or "mechanical" aids)	

The National Autism Plan For Children (NAPC)

- General developmental assessment
- Multi-agency, multi-disciplinary assessment
- Diagnosis of ASD
- Providing a baseline assessment of skills and difficulties for both the child and the family

Essential components for a complete assessment will require:

- All professionals in the multidisciplinary team involved in the diagnostic assessment of ASD require specific training
- Professionals will need to train in the use of specific assessment tools
- Does not specifically recommend which tools but mentions them

Essential components for a complete multi-agency assessment (MAA)

- 1 Existing information from all settings should be gathered.
- 2 A specific ASD developmental and family history should be taken. No evidence exists on which to recommend any particular framework, but this history should be taken by an experienced team member with recognised ASD training. In some cases it may be useful to use a semi-structured interview such as the *Autism Diagnostic Interview (ADI-R)* or the *Diagnostic Interview for Social and Communication Disorders (DISCO)*. If the person taking the developmental history is not medically trained, then the medical history and examination should be completed separately.
- 3 Focused observations should be taken across more than one setting. This could include tools such as the *Autism Diagnostic Observation Schedule (ADOS)*. The focus of the assessment of primary school aged children should include their functioning in an educational setting.
- 4 A cognitive assessment should be performed in an appropriate setting by either a clinical or an educational psychologist with ASD training.
- 5 A communication assessment should be made and speech and language competences assessed where needed by a speech and language therapist with ASD training.
- 6 An assessment should be made of mental health and behaviour. Co-morbid mental health and behaviour problems are common.
- 7 An assessment of the needs and strengths of all family members should be undertaken.
- 8 A full physical examination should be performed including appropriate medical tests.
- 9 Choice of tests will depend on each child's clinical presentation but chromosome karyotype and fragile X DNA analysis are the only current routine recommendations (Grade B). Clinical evidence of co-morbid medical conditions such as epilepsy should be sought but tests such as EEG not undertaken unless clinically appropriate. The evidence base for all investigations should be fully explained to parents.
- 10 Other assessments may be required to investigate unusual sensory responses, motor planning and co-ordination difficulties and self-care problems.

Key actions

- 1 Easy and transparent access to assessment within a specified time frame.
- 2 Discussion of the diagnosis, taking into account a sensitive framework for sharing information.
- 3 Easy access for families to information and support in relating it to their needs.
- 4 Multi-agency, multi-disciplinary assessment and working.
- 5 Appropriate intervention.
- 6 Immediate appointment of a key worker for the family.
- 7 Care plan developed with and for the family.
- 8 Care management for complex situations and ongoing needs.
- 9 Regular ASD specific training of all professionals working in assessment and provision of services.
- 10 Strategic planning and co-ordination of ASD services for local populations must be undertaken.

Diagnosis

- Diagnostic history
 - ADI
 - DISCO
 - 3Di
 - Lewisham
- Examination
- Blood tests
- ADOS
- SLT
- Early Years/nursery/school



Interviews

- Short
 - Social communication questionnaire
 - Lewisham
- Medium CHAT?
 - 3Di (computer based)
- Long All interviews subject to parental recall and bias
 - ADI-R
 - DISCO

Lewisham interview

<p>COMMUNICATION SKILLS (Concerns now or in the past) Interest in communication</p> <p>How does your child tell/show you what s/he wants? Does your child Use your hand as a tool? Get things for self? Point to request? (CHAT 18-24m) Point to share interest? (CHAT 18-24m) Follow a pointed finger? Initiate communication spontaneously? Bring toys or books to show you or share? Can your child request help? How?</p>	<p>A1 Qualitative impairment of receptive or expressive language used for social communication B1(a) failure adequately to use eye to eye gaze, facial expression, body posture and gesture to regulate social interaction B2(b) relative failure to initiate/sustain conversation (at level of language skills present), with reciprocity B1(d) lack of spontaneous seeking to share enjoyment, interests or achievements with other people (eg showing, bringing or pointing out to others objects of interest to the child)</p>
---	--

Lewisham interview

DSM-IV criteria				
6 of 12 symptoms, minimum 2 from B1, one each from B2, B3				
B1	a	b	c	d
B2	a	b	c	d
B3	a	b	c	d

Typical health district

- For an under 16 population of 55,000
 - About 120 new referrals to Child Health*
 - Majority are talking preschoolers
 - Current therapy caseload is about 300
 - Older and more high functioning tend to go to CAMHs
- Short interview, ADOS and feedback take 3 appointments, discussion with other professionals, report writing
 - Total 3.5 – 4 hours

* Over 4 times the NAPC estimate

Typical health district

- ADOS not really necessary if diagnosis is obvious and consensual
- Long interviews
 - ADI-R
 - DISCO
 - Only really practical for disagreement, second opinions and tertiary assessment
- Computer based interviews (3Di)
 - Only useful for higher functioning over 6

Investigations

- **Karyotype**
- **Fragile X**
- **CK in preschool boys**

Epilepsy:

- **EEG**
- **MRI**

Autism Diagnostic Observation Schedule

- Objective Standardised
- Structured
- Social 'presses'
- Language & communication
- Reciprocal social interaction
- Play
- Stereotyped behaviours/restricted interests
- Other abnormal behaviours

ADOS

- 4 modules based on language level
 - 1. single word
 - 2. phrase
 - 3. relationships
 - 4. independence
- Scoring
 - Autism
 - ASD
- Relates directly to ADI-R

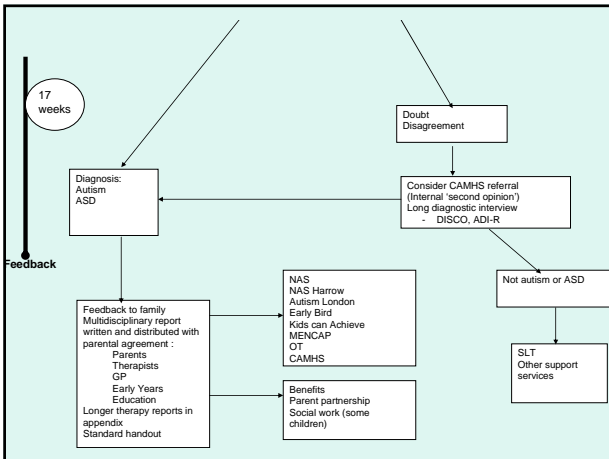
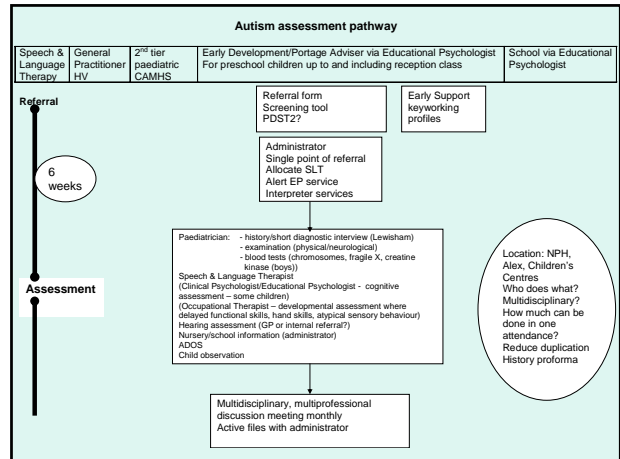
Four modules each requiring 35-40 minutes to administer (and as long again to score):

- **Module 1**
 - Free play
 - Response to name
 - Response to joint attention
 - Bubble play
 - Anticipation of a routine with objects
 - Responsive social smile
 - Functional and symbolic imitation
 - Birthday party
 - Snack

- **Module 2**
 - Construction task
 - Make-believe play
 - Joint interactive play
 - Conversation
 - Response to joint attention
 - Demonstration task
 - Description of a picture
 - Looking at a book
 - Free play
 - Response to name
 - Birthday party
 - Snack
 - Bubble play
 - Anticipation of a routine with objects

• **Module 3**

- Construction task
- Make-believe play
- Joint interactive play
- Demonstration task
- Description of a picture
- Telling a story from a book
- Cartoons
- Reporting a non-routine event/conversation
- Emotions
- Social difficulties/annoyance
- Break
- Friends/loneliness/marriage
- Creating a story



Asperger syndrome High functioning

- Australian Asperger questionnaire (Attwood)
- Gillberg and Gillberg criteria

THE AUSTRALIAN SCALE OF ASPERGER'S SYNDROME

Primary age range
Scoring: normally expected (rarely) = 0, frequently = 6

A. SOCIAL AND EMOTIONAL ABILITIES

1. Does the child lack an understanding of how to play with other children? For example, unaware of the unwritten rules of social play?

0	1	2	3	4	5	6
---	---	---	---	---	---	---
2. When free to play with other children, such as school lunchtime, does the child avoid social contact with them? For example, finds a secluded place or goes to the school library?

0	1	2	3	4	5	6
---	---	---	---	---	---	---
3. Does the child appear unaware of social conventions or codes of conduct and make inappropriate actions and comments? For example, making a personal comment to someone but the child seems unaware of how the comment could offend.

0	1	2	3	4	5	6
---	---	---	---	---	---	---

DIAGNOSTIC CRITERIA FOR ASPERGER SYNDROME (Gillberg and Gillberg 1989)

1. Severe impairments in reciprocal social interaction (at least 2 of the following):

Inability to interact with peers		
Lack of desire to interact with peers		
Lack of appreciation of social cues		
Socially and emotionally inappropriate		

2. All-absorbing narrow interests (at least one of the following):

Exclusion of other activities		
Repetitive adherence		
More rote than meaning		

3. Imposition of routines and interests (at least one of the following):

On self, in aspects of life		
On others		

4. Speech and language problems (at least 3 of the following):

Delayed development	
Superficially perfect expressive language	
Formal, pedantic language	
Odd prosody, peculiar voice characteristics	
Impairment of comprehension including misinterpretations of literal/implied meanings	

5. Nonverbal communication problems (at least one of the following):

Limited use of gestures	
Clumsy/gauche body language	
Limited facial expression	
Inappropriate expression	
Peculiar, stiff gaze	

6. Motor clumsiness, poor performance on Neurodevelopmental examination

The best way of diagnosing?

- Experience
- Experience
- Experience!

THE END

