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## Early detection of autism. Diagnostic instruments for clinicians

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**Abstract** Autism and Asperger syndrome are disorders with early childhood onset. They are believed to exist on the same spectrum of impairments of reciprocal communication and social interaction restriction of imagination and behaviour. A number of screening and diagnostic tools have been developed in the field, and several of these are briefly reviewed here. It is concluded that autism may be

screened around age 18 months and a diagnosis reliably be made around age 30 months, whereas a diagnosis of Asperger syndrome is not usually suspected, screened or made until into the child's school age.

**Key words** Autism – early diagnosis – screening questionnaires – diagnostic interviews

### Introduction

Childhood autism as defined by the ICD-10 (65) and autistic disorder as defined by the DSM-III-R (2) or the DSM-IV (3) refer to a diagnostic concept comprising the triad of restriction of reciprocal communication, reciprocal social interaction and imagination/behaviour. All three diagnostic systems presuppose infancy or childhood onset of problems, and the ICD-10 and DSM-IV require delays or abnormal functioning prior to age 3 years. Autism is associated with mental retardation in about 65–90% of the cases (61) and few individuals have a full-scale IQ above 80. Given the general agreement that most, but not all, cases with severe triad symptoms commence in the first few years of life, researchers and clinicians should be able to establish the diagnosis in early childhood.

Asperger syndrome (3, 23, 54, 59, 65), also comprises triad symptoms, albeit usually of a milder degree. Most authorities regard it as a variant of “high-functioning autism” (21). Asperger syndrome is now often grouped with autistic disorders and atypical autism/autisticlike conditions (and other “triad” conditions) under the blanket

term of autism spectrum disorders (27, 53, 60). Being generally associated with higher levels of IQ than autism (few individuals with this diagnosis have tested IQ:s under 80), obvious symptoms are sometimes not recognized until well into the child's school age.

Even though a preliminary diagnosis of autism spectrum disorder (22, 53) or Asperger syndrome may well be made in the first 5 years of life, the typical clinical gestalt often only emerges (or rather, is recognized) in the 6–10-year-old age range.

Several different screening and diagnostic instruments for autism/autism spectrum disorders have been developed over the last 20 years. For these instruments (as for clinical diagnostic work), one important factor is the delineation of autism spectrum problems from the effects of global developmental retardation.

This paper provides a brief review of the instruments (Table 1) and concludes by listing some general clinical recommendations.

**Table 1** Screening and diagnostic instruments in the field of autism and Asperger syndrome

Instrument	Age range	Reference
<i>Screening:</i>		
<i>autism</i>		
SAB-2	0–2 years (or retrospective version)	Dahlgren et al., 1989
CHAT	1.5–3 years	Baron-Cohen et al., 1992
ABC	3 years plus	Krug et al., 1980
<i>Asperger syndrome</i>		
ASSQ	7–16 years	Ehlers & Gillberg, 1993
<i>Diagnosis and symptom rating:</i>		
<i>autism</i>		
IBSE	0.5–4 years	Barthélémy et al., 1993
CARS	all ages	Schopler et al., 1988
HBS	all ages	Wing & Gould, 1978
DISCO	all ages	Wing & Gould, 1994
ADI-R	from MA* 18 mo.	LeCouteur et al., 1989, Lord et al., 1994
ADOS	6–18 years	Lord et al., 1989
PL-ADOS	3–6 years	DiLavore et al., 1995
BSE	all ages	Barthélémy et al., 1990
RLRS	all ages	Freeman et al., 1986
ABI	all ages	Cohen et al., 1993
<i>Asperger syndrome</i>		
ASDI	7–16 years	Gillberg et al., 1994

\* MA = mental age

## Autism screening and diagnosis

### Findings in the newborn period and the first year of life

No instruments can reliably detect the syndrome of autism in the first 6–12 months of life. In a study of infant screening-test records, children with subsequently diagnosed autism had lower incidence of problems at 12 months than a control group with non-specific developmental delay. At 18 months, children with autism had a selective increase in abnormalities of social interaction (33). Analysis of home videotapes have indicated that impairments in both social and joint attention behaviours can be found at the age of 1 year (43).

Autism is one of the syndromes to be kept in mind in all follow-up of children with early signs of developmental delay and/or neurologic disorder. Genetic factors are important in many cases of autism (9), either without other signs of neurodevelopmental problems or coexisting with certain well-known hereditary neurologic disorders, e.g. tuberous sclerosis (28, 31). Also many clear non-genetic causes exist, e.g. congenital rubella (11).

Specific covariation have been found between autism and infantile spasms (45). Also, other epileptic syndromes may carry an increased risk of autism, especially syndromes that start early in life and are combined with mental retardation (51).

Muscular hypotonia (including “floppy infant syndrome”) is a common antecedent of developmental disorders including autism. For instance, the fragile X syndrome, one hereditary cause of mental retardation and autism, is often associated with muscular hypotonia (32).

Studies of total populations of children with mental retardation show high prevalences of autism and autistic-like conditions (20–23% in severe mental retardation and 5–6% in mild mental retardation, (30, 41). It is not possible to reliably separate the “core” hereditary cases from so-called “symptomatic” cases only by behavioural characteristics. Also among siblings with autism there is a high intrafamilial variability in symptoms and IQ (50).

Minor physical anomalies may give hints of a prenatal origin of a disorder (damage to the fetus and/or genetic factors). Three independent groups of researchers have documented the presence of certain minor physical anomalies in autism. Ear anomalies have been among the most commonly reported findings, including low-set ears (which could contribute to a high rate of ear infections and conductive hearing loss in autism (49), adherent ear lobes, asymmetrical, soft and pliable ears and malformations of the auricle (10, 37, 58). Other physical anomalies reported to be associated with autism include hypertelorism, partial syndactyly of the second and third toes and abnormalities of the mouth (high-arched palate, tongue furrows and smooth/rough spots).

**Parental interview and behavioural observation aiming at detection of autism in the first 18–24 months of life**

SAB-2-checklist

(symptoms of autism before age 2 years)

In a series of studies from Göteborg, Sweden, early symptoms of autism have been examined by both retrospective, current and prospective study (14, 24, 25). In the first of these, a 130-item questionnaire (a visual analogue scale) referring to symptoms of autism under age 2 years was completed by mothers of 7–22-year-old children with autism recruited after population screening and by mothers of sex- and age-matched normal children and sex- and age-matched children with mental retardation. The instrument showed good discriminatory capacity for autism

vis-à-vis mental retardation and normality. The results of this retrospective study must be interpreted with caution because of possible bias that may be associated with selective recall, memory distortion and failure to observe the pertinent signs of autism. Because mother reports were used, interrater reliability could not be tested. Furthermore, test-retest reliability of the questionnaire has not been properly established. In the second study, the same questionnaire was used in a prospective investigation of children with autism and related disorders who were seen before age 3 years. The results were contrasted with findings obtained in an age-, sex- and IQ-matched comparison group without autistic symptoms. Table 2 lists the 28 items which characterized the autism group in either the retrospective study only (8 items), the prospective study only (9 items), or both studies (11 items).

**Table 2** Items discriminating autism from mental retardation and normality under age 3 years

Area/item	Prospective study (Gillberg et al., 1990) v = prospective study only a = retrospective study also	Retrospective study (Dahlgren et al., 1989) v = retrospective study only a = prospective study also
<i>Social</i>		
Appears to be isolated from surroundings	a*	a
Doesn't smile when expected to	a	a
Difficulties getting eye contact	a	a
Doesn't matter much whether Mom or Dad is close by or not	v	
Doesn't like to be disturbed in own world		v
Content if left alone		v
<i>Communication</i>		
Doesn't try to attract adult's attention to own activity	a	a**
Difficulties imitating movements	a	a
Late speech development	v	
Doesn't point to objects	v	
Doesn't understand what people say	v	
Can't indicate own wishes	v	
<i>Play behaviour</i>		
Doesn't play like other children	a*	a
Occupies her/himself only when alone	a	a
Plays only with hard objects	a	a
Odd attachments to odd objects		v
<i>Perception</i>		
There is (or has been) a suspicion of deafness	a*	a
Empty gaze	a	a**
Overexcited when tickled	a	a
There is something strange about his/her gaze	v	
Interested only in certain parts of objects	v	
Exceptionally interested in things that move	v	
Doesn't listen when spoken to	v	
Strange reactions to sound		v**
Doesn't seem to react to cold		v
Engages in bizarre looking at objects, pattern and movements		v
<i>Rhythmicity</i>		
There are days/periods when s/he seems much worse than usual		v
Severe problems over sleep		v

\* Three items with strongest discriminatory power in prospective study

\*\* Three items with strongest discriminatory power in retrospective study

a = applies

On the basis of the results obtained in these studies and long-term clinical experience with infants, young children and adolescents and adults with autism, Gillberg and Coleman (24) suggested an observational screening device for autism in the 10–18 months age range (Table 3).

#### IBSE (Infant Behavioural Summarized Evaluation scale)

This observation scale has been adapted from the BSE (Behavioural Summarized Evaluation scale; see below) for use with infants 6–48 months of age. A 19-item version is extracted from the initial experimental scale (1). Inter-rater reliability was reported to be acceptable in the original study by the developers of the IBSE. It has the particular advantage of having a companion version for older children. It is neither a pure screening instrument nor a diagnostic scale. It appears to be intended for the quantified recording over time of behaviours typically associated with autism in very young children.

#### CHAT (Checklist for Autism in Toddlers)

In a preliminary study of siblings of children with autism (5), this brief checklist was shown to have good discriminating capacity screening for autism. Test-retest and inter-rater reliability have been reported to be acceptable to good. Only children who later developed the full syn-

**Table 3** Screening for autism at the well-baby clinic at ages 10 and 18 months

1) The following questions to the mother provide a tentative framework for a check-list to be used whenever there is (even mild) suspicion of autistic-like behavior or autism:

- Do you consider your child's eye-to-eye-contact to be normal?
- Do you think that s/he listens to you or has normal hearing, or does s/he react only to particular sounds?
- If there are or have been any feeding problems or abnormal behaviors in connection with feeding, what were they?
- Is s/he comforted by proximity or body contact?
- Does s/he oppose body contact?
- Does s/he show any interest in his/her surroundings?
- Does s/he often smile or laugh quite unexpectedly?
- Does s/he prefer to be left alone?
- Is your child, on the whole, like other children?

2) Examine the following features systematically:

- Hand stereotypes (including strange looking at or posturing of hands)
- Avoidance of gaze contact
- Stiff, staring gaze
- Rejection of body contact
- No or very variable reaction to strong, unexpected noise
- Obvious lack of interest (e.g. does not show interest in peck-a-boo games)

drome of autism had abnormal scores on this instrument around age 18 months. In a more recent study, this instrument was used in a population of 16000 children who were not severely mentally retarded. Again, this instrument was shown to be useful as a screening tool with a low rate of false positives (4, 52).

#### Pre-school age and later

At the present, autism is a diagnosis which should not be made with certainty until the child is at least 30–36 months of age. A tentative diagnosis can usually be made much earlier.

#### CARS (Childhood Autism Rating Scale)

The CARS (47) is still, without doubt, the best validated instrument for diagnosis along the autism spectrum (44, 48), and has been in widespread use for the last 15 years. It is based on behavioural observation and/or interview. It has acceptable or good psychometric properties among child, adolescent and adult populations of developmentally disordered individuals. It covers 14 domains generally affected by severe problems in autism, plus an overall category of "impression of autism". Each possible problem area is rated on a 1–4 scale (1 indicating behaviour appropriate for age and 4 indicating severe abnormality) with use also of midpoints between adjacent scorings (as 1,5) giving a continuum of 7 points. Scores of 29 or under are considered indicative of "non-autism", whereas scores of 30–36 are believed to reflect "mild or moderate autism" and scores of 37–60 "severe autism".

The CARS should not be used as the only instrument for deciding on a diagnosis of autism, but it is definitely useful as an aid in diagnosis and as a guide to the severity of the syndrome, particularly in children who have mental retardation as well as autism (to our knowledge no study of the CARS validity has been performed on a population with autism and normal IQ). The results of the CARS (as of any other scoring list) must be analyzed with extra care in children with severe or multiple other dysfunctions. Disorders of mobility, vision and/or hearing may yield high rates of false negatives.

#### HBS (Handicaps, Behaviour and Skills schedule)

This semi-structured, clinical and investigator-based interview was developed to cover the whole range of impairments in social interaction, communication and symbolic play usually found in the syndrome of autism (62). It has been used for more than 15 years. Reliability (inter-and intra-rater) is good according to several different studies (8, 42, 62). It has been shown to be useful in studies of children and adults alike (40, 63).

The HBS is not regarded as a diagnostic instrument specific to narrow autism. It aims to elicit the information necessary to arrive at diagnoses on the autism spectrum and it is particularly geared to the study of individuals who also suffer from mental retardation. The interview gathers information on the child's functioning and behaviour in daily life situations. The Vineland Social Maturity Scale (16) can be completed from the information elicited. The internal consistency and content validity of the developmental score were found to be satisfactory in a recent study (55). The HBS has many merits, including its explicit flexible use depending on other disorders and on the overall intellectual functioning of the individual. It is now available in a recently updated version, the DISCO (Schedule for Diagnosis of Social and Communication Disorders) (64).

#### ADI-R (Autism Diagnostic Interview-Revised)

The ADI is a standardized, semistructured and investigator-based interview, which was shown to effectively discriminate between autistic and non-autistic mentally handicapped individuals (35). Its reliability was good or excellent. The original ADI was validated for children from the age of 5 years with a mental age  $\geq 2$  years. The revised version, ADI-R, has been modified to be appropriate for children with mental ages from about 18 months. Good psychometric data have been presented for a sample of pre-school children (38).

The interview aims to assess the lifetime development in the areas of interest for autism diagnosis and to clearly separate qualitative deviations from delay of development. Many ratings are focused on the 4 to 5 years age period, when certain diagnostic features are thought to be most evident. An algorithm is linked to ICD-10 and DSM-IV criteria.

The interview is designed to be administered to the subject's principal care-giver by a trained interviewer. The task of the interviewer is to elicit descriptions of behaviour and make codings according to defined concepts for every item. The procedure is time-consuming. The ADI is currently one of the most used instruments in research on autism. As such it is excellent for recruiting nuclear autism cases, but appears to have less import in the study of problems on the brighter end of the autism spectrum, including Asperger syndrome. The current version of the ADI does not include items concerning verbal comprehension.

#### ADOS (Autism Diagnostic Observation Schedule)

The ADOS (39) is the first available structured observation schedule specifically developed for use in the diagnosis of autism. The ADOS is designed to focus on the *qual-*

*ity* of social and communicative behaviour. It is an *interactive* instrument using a standardized set of social contexts (with standardization of the interviewer's behaviour, tasks and material). It aims to encourage interaction that appears natural and to use prompts when necessary. The ADOS is designed for subjects with an estimated mental age of 3 years or greater. In the preliminary study it was used to distinguish individuals with and without autism, who were 6–18 years of age and had normal IQ or mild mental retardation. The first preliminary study showed good psychometric properties (reliability and validity).

An algorithm is developed for the ADOS (like for the ADI) to give a diagnosis according to ICD-10 criteria. The authors emphasize that the ADOS does not always give adequate information for assessment of the quality of behaviour and restriction of interests. Most children with autistic problems benefit so much from structure (12) that their most severe and obvious symptoms will not be shown in an environment with a high degree of structure. Therefore, information from other sources is needed, e.g., from observation in unstructured settings and from parent interviews.

The validity of this instrument is highly dependent on the training and clinical skills of the examiner. The ADOS is limited to use among more able children with developmental disorders. The same research group has now developed an instrument for younger/non-verbal subjects, employing the same principles and general strategies as the ADOS, but with a format which is more flexible with briefer activities, allowing the child to move around the examination room. This instrument is referred to as the Pre-Linguistic ADOS or PL-ADOS (15).

#### ABC (Autism Behaviour Checklist)

The ABC is a 57-item checklist first developed as a screening instrument for autistic behaviour in populations with mental retardation (34). It has since been widely used in populations of children, adolescents and adults with autism (34, 56, 57). It is generally recognised as a reasonable tool for estimating the degree of autistic symptomatology in an individual and for following up such symptoms over time. However, concern about its reliability (44, 56) and specific diagnostic validity – at least if the cut-off for autism suggested by the original researchers is adhered to (41, 48, 57) – has precluded its general use both as a screening tool and diagnostic instrument. A recent study has shown reliable information when using retrospective parental ABC ratings (symptoms at 3–5 years) but little diagnostic help from current ratings in a group of children with autism and normal intellectual abilities (66). The checklist might have its focus more on maladaptive behaviours than on the lack of social and communicative skills (48). One problem (that is common to many other checklists, see above regarding

the CARS) is the difficulties using the instrument in heterogeneous populations with various forms of dysfunction (making it impossible to respond to many items).

There is some support that a 17-item questionnaire, comprising only the most highly weighted items of the 57-item version of the ABC, may be just as useful as the original version (19, 57).

#### BSE (Behavioural Summarized Evaluation)

This instrument, developed by a French group (36) and recently validated (6, 7) covers 20 areas related to autism symptomatology. Ratings are performed after observation of the child. The BSE is quick and simple to administer. Reliability has been reported to be acceptable or good in the French studies. It is intended for use with children of pre-school and school age. It was not developed as a diagnostic instrument, but provides a means for systematic recording of data relating to autism symptoms and for validating the effects of therapeutic interventions.

#### RLRS (Real Life Rating Scale)

This scale, developed by Freeman et al. (20), has been used in several drug studies of autism (18, 46) to follow the development of symptoms typical of autism. It is not intended for screening or diagnosis per se but is helpful in evaluating the type and severity of symptoms involved in the syndrome of autism (48). Its reliability appears to be acceptable (20).

#### ABI (Autism Behaviour Interview)

This recently developed instrument is a structured parent interview (13). The authors found satisfactory reliability and internal consistency measures for most of the subscales. They described encouraging results when using the information from the ABI in a neural network model, a computer program that resembles the parallel processing of the nervous system. The diagnostic process might be more like recognition of a non-linear pattern than summing up numbers of items/behaviour descriptors as in traditional diagnostic schedules.

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### Asperger syndrome screening and diagnosis

Asperger syndrome is only rarely picked out by any of the aforementioned instruments, except in cases with early onset symptomatology suggestive of full-blown autistic disorder (that later develops into more typical Asperger syndrome symptoms). Also, the syndrome is rarely clinically striking until age 5–6 years and above, even though abnormalities of communication and social interaction

(and behaviour) have usually been present long before this age even when obvious symptoms have been subtle.

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### Screening and diagnostic instruments

Just recently, a few screening and diagnostic aids have been developed in this (fast expanding) field of autism spectrum disorder research.

#### ASSQ (Asperger Syndrome Screening Questionnaire)

In a study by Ehlers and Gillberg (17) a teacher questionnaire containing 27 items was used in screening for Asperger syndrome. This instrument was developed in a joint venture between Lorna Wing and the Swedish group. It has good reliability (across raters and time) and appears to be useful in screening social deficits on the autism spectrum. However, with a suggested cut-off score of 5 or more (possible range 0–54), not only Asperger syndrome but other cases with social abnormalities are identified. A recent study in a clinical setting showed 13 to be the best cut-off for parents and 22 for teachers (Ehlers, unpublished data).

#### ASDI (Asperger Syndrome Diagnostic Interview)

The ASDI is currently only available in a parent version intended for diagnosis after interview with a parent (or other caregiver who knows the child well) (26). It contains questions relating to the 20 areas of possible dysfunction comprised in the diagnostic criteria outlined by Gillberg and Gillberg (27) and elaborated in Gillberg (23). Its discriminative validity within the autism spectrum disorders group remains to be established, but it seems clear that it differentiates Asperger syndrome from normality (17). A diagnostic algorithm is provided.

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### Conclusions and clinical recommendations

The clinician should aim for autism diagnosis in the pre-school period. The diagnosis of Asperger syndrome may have to be postponed until school age in many cases. There are a number of screening instruments that can be used. It appears that the CHAT (for autism in toddlers) and the ASSQ (for Asperger syndrome in school age children) may hold particular promise and may be useful in screening for autism and Asperger syndrome in well-baby clinics and schools. Neither takes more than about 5 min to complete. Cut-off scores for possible "cases" have been suggested by the authors of the original publications.

In clinical practice, diagnosis can be aided by the use of the CARS and the HBS/DISCO in the field of autism

and, perhaps, by the ASDI in Asperger syndrome. These instruments can be used flexibly and require 30–120 min to complete. The ADI and ADOS are detailed clinical and research instruments for which prior training is needed. The ABC, the RLRS, the IBSE and BSE may be helpful in monitoring change over time in people with autism. Further studies are needed concerning use of the ABI and neural networks.

Finally, it should be added that screening instruments, rating scales and diagnostic interviews cannot replace the taking of a detailed developmental history and clinical examination by an examiner experienced and skilled in the field of autism and Asperger syndrome. Also, once a tentative diagnosis has been made, there is a need to assess the problem areas and skills in the individual child and to perform a comprehensive medical work-up (24).

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