Autism Spectrum Disorder (ASD) — Revision Sheet

Supervised by Dr. Amir Naghshzan (AmirNaghshzan@gmail.com)

1. Definition

Autism spectrum disorder (ASD) is a neurodevelopmental disorder characterized by persistent difficulties in **social communication and social interaction**, together with **restricted, repetitive patterns of behavior, interests, or activities**.

Symptoms begin in the developmental period and cause clinically significant impairment in social, educational, occupational, or adaptive functioning. Presentation is highly variable. (Nice)

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2. Clinical Presentation

A. Two core diagnostic domains

1. Social communication and interaction

Look for:

- Reduced or abnormal **eye contact**
- Reduced response to name
- Poor **joint attention**
  - e.g., does not point to show something interesting
- Difficulty sharing interests or emotions
- Reduced reciprocal conversation
- Difficulty understanding social cues
- Difficulty developing peer relationships
- Limited pretend or imaginative play
- Unusual social approaches or poor reciprocity

2. Restricted/repetitive behaviors or interests

Examples:

- Repetitive movements:

    - hand flapping
    - rocking
    - spinning
- Repetitive speech:
    - echolalia
    - repeating phrases
- Insistence on sameness/routines
- Distress with minor changes
- Highly restricted or intense interests
- Sensory abnormalities:
    - hypersensitivity to sound
    - unusual response to textures
    - fascination with lights/movement
    - unusual sensory seeking

B. Important developmental clues

Ask specifically about:

- Language development
- Social smile
- Response to name
- Pointing and joint attention
- Imitation
- Pretend play
- Interaction with other children
- Loss of previously acquired skills

🚩 Developmental regression

Loss of previously acquired language or social skills is a major red flag.

Regression does **not** exclude ASD, but it should prompt careful assessment for other neurological and medical causes.

Particularly important:

Motor regression → neurological assessment.

NICE recommends pediatric/neurologic assessment for children with motor regression and referral pathways for significant language/social regression. (Nice)

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3. Diagnostic Approach

The practical algorithm

Developmental concern

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Step 1 — Developmental surveillance

Ask:

- Is development appropriate?
- Are there concerns from parents/teachers?
- Is there regression?
- Are social communication difficulties present?

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Step 2 — Developmental screening

AAP recommends:

- General developmental screening: **9, 18 and 30 months**
- ASD-specific screening: **18 and 24 months**

Screen earlier/later whenever there is clinical concern. (CDC)

A commonly used ASD screening instrument is:

M-CHAT-R/F for toddlers.

Important:

Screening ≠ diagnosis.

A positive screening test means:

"This child needs further evaluation."

It does **not** mean:

"This child has autism."

(CDC)

Step 3 — Comprehensive clinical assessment

Diagnosis should integrate:

A. Parent/caregiver history

Ask about:

- Developmental milestones
- Language
- Social interaction
- Play
- Repetitive behaviors
- Sensory abnormalities
- Adaptive functioning
- Regression
- School/daycare functioning
- Family history
- Pregnancy/perinatal history

B. Direct observation

Observe:

- Eye contact
- Response to name
- Joint attention
- Social reciprocity
- Communication
- Play
- Repetitive behaviors
- Flexibility with changes

C. Assessment of developmental domains

Assess:

- Cognitive ability
- Language
- Motor function
- Adaptive functioning

- Social-emotional development

The AAP emphasizes assessment across multiple developmental domains rather than focusing only on autism-specific symptoms. (American Academy of Pediatrics)

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4. Differential Diagnosis

Condition	Important distinguishing feature
Developmental language disorder	Language impairment without the characteristic restricted/repetitive behaviors of ASD
Intellectual disability	Global developmental impairment; social communication difficulties may occur but are generally consistent with developmental level
Hearing impairment	Poor response to name/communication difficulties; hearing assessment is essential
ADHD	Inattention/hyperactivity/impulsivity rather than primary social-communication deficits
Social (pragmatic) communication disorder	Social communication impairment without restricted/repetitive behaviors
Rett syndrome	Regression with characteristic neurological phenotype; especially important in girls
Childhood-onset schizophrenia/psychosis	Later onset with psychotic symptoms rather than early developmental social-communication abnormalities
Reactive attachment disorder	Abnormal social interaction associated with severe neglect/adversity
Selective mutism	Child may communicate normally in some settings but not others
Anxiety disorders	Social avoidance may mimic ASD but developmental history is different

Examination pearl

Language delay alone ≠ autism.

The presence of **restricted/repetitive behaviors or interests** is crucial to the diagnosis of ASD.

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5. Investigations

There is NO routine laboratory test for diagnosing ASD.

Diagnosis is clinical. (CDC)

Recommended assessment

Usually appropriate:

- Hearing assessment
- Vision assessment
- Developmental/cognitive assessment
- Speech and language assessment
- Adaptive functioning assessment

Etiologic evaluation

Consider **genetic evaluation**, particularly after diagnosis.

AAP recommends offering:

- **Chromosomal microarray**
- **Fragile X testing**

with additional genetic testing guided by phenotype/family history and specialist assessment. (American Academy of Pediatrics)

Do NOT routinely order:

- Brain MRI
- EEG
- Metabolic testing
- Extensive biochemical investigations

These should be **clinically indicated**, not performed routinely simply because a child has ASD. (American Academy of Pediatrics)

When EEG/MRI becomes important

Consider when there are:

- Seizures
- Abnormal neurological examination
- Significant neurological regression
- Focal neurological findings
- Other specific neurological concerns

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6. Management Algorithm

ASD suspected

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1. Do not wait for definitive diagnosis

If developmental delay is identified:

Start appropriate developmental intervention while diagnostic assessment is ongoing.

(American Academy of Pediatrics)

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2. Comprehensive developmental assessment

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3. Confirm ASD / identify coexisting conditions

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4. Individualized intervention

Main components:

A. Behavioral/developmental intervention

- Structured behavioral interventions
- Communication-focused interventions
- Social skills interventions
- Parent-mediated interventions
- Educational interventions

B. Speech and language therapy

Especially when there is:

- Language delay
- Poor functional communication
- Social communication difficulties

C. Occupational therapy

May help address:

- Functional skills
- Adaptive skills
- Sensory-related difficulties

D. Educational support

Individualized according to developmental and educational needs.

5. Identify and treat comorbidities

Screen for:

- ADHD
- Anxiety/depression
- Epilepsy
- Sleep disorders
- Feeding difficulties
- Gastrointestinal symptoms
- Intellectual disability
- Obesity
- Wandering/elopement
- Challenging behavior

These comorbidities frequently determine the child's clinical needs. (American Academy of Pediatrics)

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7. Drug Summary Table

No medication treats the core symptoms of ASD.

Problem	Management
Core social-communication deficits	**Behavioral/developmental intervention—not medication**
Repetitive behaviors	Behavioral intervention
ADHD symptoms	Standard ADHD treatment may be considered after assessment
Anxiety/depression	Psychological/psychiatric assessment and treatment when indicated

Severe irritability/aggression	Specialist assessment; antipsychotic medication may be considered
Sleep problems	Behavioral sleep interventions first; medication only when appropriate

Antipsychotics

Risperidone and aripiprazole may be used for **severe irritability/aggression/self-injurious behavior** associated with ASD in appropriate age groups and under specialist supervision.

They **do not treat the core features of autism**.

Monitor for:

- Weight gain
- Metabolic effects
- Extrapyramidal effects
- Sedation

NICE specifically recommends that medication for challenging behavior be considered only after assessment of triggers and psychosocial/environmental interventions, with careful monitoring. (Nice)

🚫 Do not use medications to "cure" autism

NICE recommends against using:

- Antipsychotics for core ASD symptoms
- Antidepressants for core ASD symptoms
- Anticonvulsants for core ASD symptoms
- Gluten-free/casein-free exclusion diets for core ASD symptoms

(Nice)

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8. Admission Criteria

ASD itself is generally NOT an indication for hospital admission.

Admission may be necessary because of a **coexisting acute problem**, such as:

- Severe self-injury
- Severe aggression with immediate safety risk
- Acute psychiatric crisis

- Severe dehydration due to feeding refusal
- Status epilepticus/seizures
- Serious injury
- Acute medical illness
- Severe behavioral crisis that cannot safely be managed in the community

Emergency principle

In an acutely agitated autistic child, **look for pain, illness, sensory overload, communication difficulty, medication effects, or environmental triggers before assuming the behavior is "just autism."**

NICE emphasizes assessment for physical illness, mental health problems, environmental factors and changes in routine when challenging behavior occurs. (Nice)

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9. Referral Criteria

Refer for specialist developmental/autism assessment when:

- ASD is suspected clinically
- Screening is positive
- Significant social-communication abnormalities are present
- Restricted/repetitive behaviors are present
- Developmental concerns persist despite an initially negative screen
- There is developmental regression
- There is diagnostic uncertainty

NICE recommends referral based on clinical concern and emphasizes that autism should not be excluded simply because a child does not display every typical feature. (Nice)

Additional referrals

Finding	Referral
Speech/language delay	Speech & language therapy
Hearing concern	Audiology
Seizures	Neurology
Significant developmental delay	Developmental pediatrics
Genetic phenotype/family history	Clinical genetics
Severe behavioral problems	Behavioral/mental health specialist
ADHD/anxiety	Appropriate mental-health/developmental service
Feeding disorder	Feeding/GI/nutrition team

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10. Follow-up

ASD requires **long-term, multidisciplinary follow-up**.

At follow-up assess:

Development

- Language
- Communication
- Cognitive development
- Adaptive skills
- School performance

Medical

- Growth/BMI
- Sleep
- Feeding
- GI symptoms
- Seizures
- Medication adverse effects

Behavioral

- Anxiety
- ADHD symptoms
- Challenging behavior
- Self-injury
- Wandering/elopement

Family

- Caregiver stress
- Educational needs
- Access to services
- Family support

NICE recommends coordinated multidisciplinary care and attention to physical, mental-health, educational and social needs. (Nice)

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11. Clinical Pearls ⭐

Pearl 1

ASD is a clinical diagnosis.

There is no diagnostic blood test or MRI.

Pearl 2

Screening is not diagnosis.

M-CHAT-R/F identifies children who need further evaluation.

Pearl 3

Normal hearing must be considered in every child with language delay.

Pearl 4

Language delay alone does not establish ASD.

Look for the combination of:

Social communication impairment + restricted/repetitive behaviors.

Pearl 5

A negative screening test does not completely exclude ASD.

Persistent clinical concern → reassess/refer. (CDC)

Pearl 6

Do not delay intervention while waiting for an ASD diagnosis.

Developmental services can begin when developmental delays are identified. (American Academy of Pediatrics)

Pearl 7

Regression is a red flag.

Especially investigate neurological causes when there is motor regression, seizures, or abnormal neurological examination.

Pearl 8

Girls may be under-recognized.

NICE specifically highlights the possibility of underdiagnosis in girls. (Nice)

Pearl 9

Treat the child, not the diagnosis.

Management should target the child's specific developmental difficulties and comorbidities.

Pearl 10

Behavioral deterioration should trigger a search for a medical cause.

Pain, constipation, sleep problems, seizures, anxiety, environmental changes and communication difficulties may present as "challenging behavior." (Nice)

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12. Common Mistakes in Exams and Practice

❌ Mistake 1

"A positive M-CHAT means the child has autism."

✅ False. It means **further evaluation is required**.

❌ Mistake 2

"Autism can be diagnosed by MRI/EEG/genetic testing."

✅ False. These tests may identify associated conditions or etiology but do not establish ASD.

❌ Mistake 3

"A child with speech delay has autism."

✅ False. Consider hearing impairment, developmental language disorder, intellectual disability and other causes.

❌ Mistake 4

"Wait until age 3–4 years before evaluation."

✅ Incorrect. ASD can be recognized and diagnosed much earlier, and early intervention should not be delayed. (CDC)

❌ Mistake 5

"Antipsychotics treat autism."

✅ They may reduce **severe irritability/aggression**, but they do not treat the core social-communication deficits.

❌ Mistake 6

"Gluten-free diet is standard treatment for ASD."

✅ No. Routine exclusion diets are not recommended for treating core ASD symptoms. (Nice)

❌ Mistake 7

Ignoring regression

✅ Regression requires careful developmental and, when appropriate, neurological evaluation.

❌ Mistake 8

Assuming all autistic children have intellectual disability.

✅ ASD occurs across a wide range of intellectual abilities.

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13. Key Take-Home Messages

1. **ASD is a neurodevelopmental disorder involving social communication/interaction deficits and restricted/repetitive behaviors.**
2. **Diagnosis is clinical**, based on developmental history and behavioral observation.
3. **Screening ≠ diagnosis.**
4. Routine ASD-specific screening is recommended at **18 and 24 months**, while developmental surveillance should continue throughout childhood. (CDC)
5. **Language delay alone does not equal ASD.**
6. Always consider **hearing impairment and other developmental disorders**.
7. **Do not wait for definitive diagnosis before starting appropriate developmental intervention.**
8. There is **no medication that treats the core symptoms of ASD**.
9. Treat important **comorbidities** such as ADHD, anxiety, epilepsy, sleep and feeding problems.
10. Severe aggression/self-injury may require specialist management and, in selected cases, medication such as **risperidone or aripiprazole**.
11. **Regression, seizures, abnormal neurological findings, or acute behavioral deterioration are red flags.**
12. ASD requires **long-term, individualized, multidisciplinary care**.

One-Minute GP Algorithm

Developmental concern?

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Check milestones + developmental history

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ASD-specific screening when appropriate (18–24 months)

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Positive screen OR persistent clinical concern

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Comprehensive developmental/ASD assessment + hearing assessment

ASD confirmed?

YES → Early intervention + assess comorbidities + family/educational support + follow-up

NO/UNCERTAIN → Reassess development + alternative diagnoses + specialist referral if concern persists

The most important clinical rule:

Do not wait for a diagnostic label before helping a child with an obvious developmental delay.

This approach is consistent with the AAP clinical report and current NICE/CDC recommendations. (American Academy of Pediatrics)

Key references

- **AAP:** Hyman SL, et al. *Identification, Evaluation, and Management of Children With Autism Spectrum Disorder.* Pediatrics. 2020. (American Academy of Pediatrics)
- **NICE CG128:** Autism spectrum disorder in under 19s—recognition, referral and diagnosis. (Nice)
- **NICE CG170:** Autism spectrum disorder in under 19s—support and management; surveillance reviewed through 2025. (Nice)
- **CDC:** Current clinical screening and diagnostic guidance for ASD. (CDC)