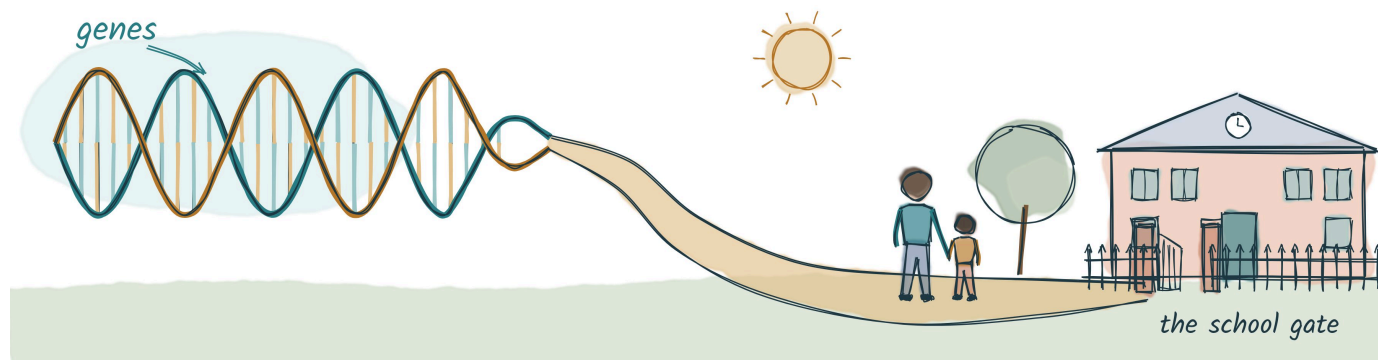


Handout — Dr Mark Aszkenasy, Consultant Paediatrician

For delegates. Companion to two plenaries — not a full transcript. Local pathways vary.



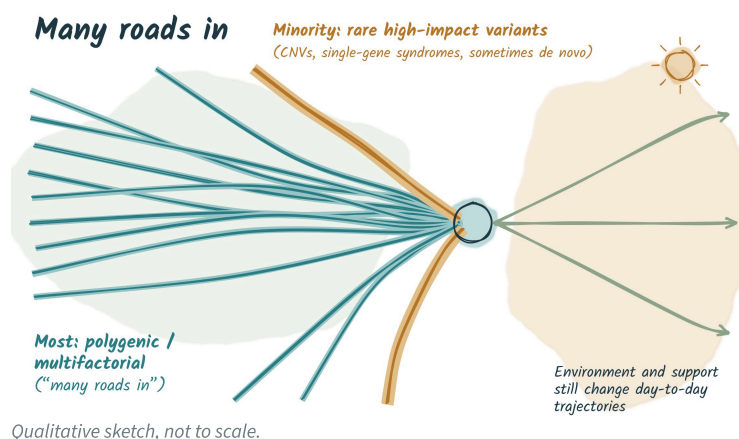
1 Opening (09:30–10:10) — The Genetic Landscape of Autism: What We Know, What to Do

Three take-homes

- Autism is highly heritable and usually not a single-gene disease.
- A normal genetic test does not mean “not genetic.”
- Test when the answer would change management, surveillance, or family counselling — not to “prove” autism.

Landscape

- Strong genetic contribution; most cases polygenic/multifactorial (“many roads in”).
- Minority: rare high-impact variants (CNVs, single-gene syndromes, sometimes de novo).
- Heritable ≠ simple inheritance; broader autism phenotype can run in families.
- Biology is real; environment and support still change day-to-day trajectories.



Qualitative sketch, not to scale.

What tests can / cannot do



Can: find a syndrome; guide surveillance; inform recurrence risk; end a diagnostic odyssey.



Cannot: binary lab “proof” of autism; explain every profile; guarantee treatment.

- Normal microarray/panel/exome still compatible with genetic autism (esp. polygenic).
- VUS needs genetics counselling — not internet interpretation.

Higher yield patterns · Poor reasons to test

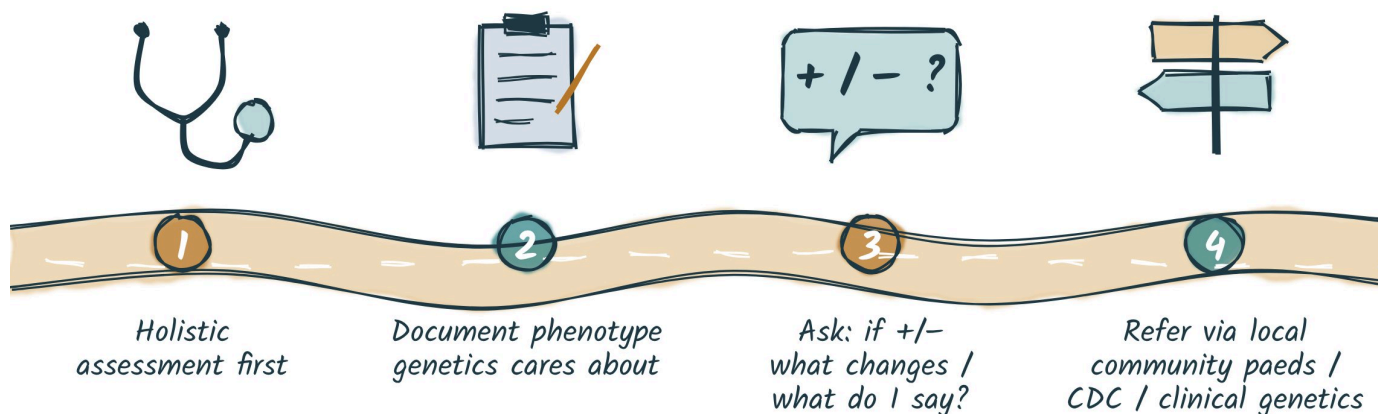


Higher yield often: ID/GDD; anomalies/dysmorphism; micro/macrocephaly; epilepsy/complex neurology; consanguinity/striking FH; multi-system disease.



Poor reasons: prove school wrong; replace developmental assessment; gene name as only goal; expect precision Rx for typical autism from one test.

Monday-morning path



Testing must not delay support, adjustments, or clear description of need

- Holistic assessment first (development, hearing/vision, sleep, gut/pain, seizures, nutrition, safeguarding).
- Document phenotype genetics cares about → ask: if +/- what changes / what do I say?
- Refer via local community paed's / CDC / clinical genetics; don't freestyle WGS outside pathway.
- Testing must not delay support, adjustments, or clear description of need (e.g. EHCP-relevant).

“ ” Family language (examples)

“Autism often runs in families in complicated ways – that doesn't mean anyone did something wrong.”

“We're not using a test to prove autism; for some children we're looking for a medical explanation that changes care.”

“A normal result is common and doesn't mean the child's differences aren't real or partly genetic.”

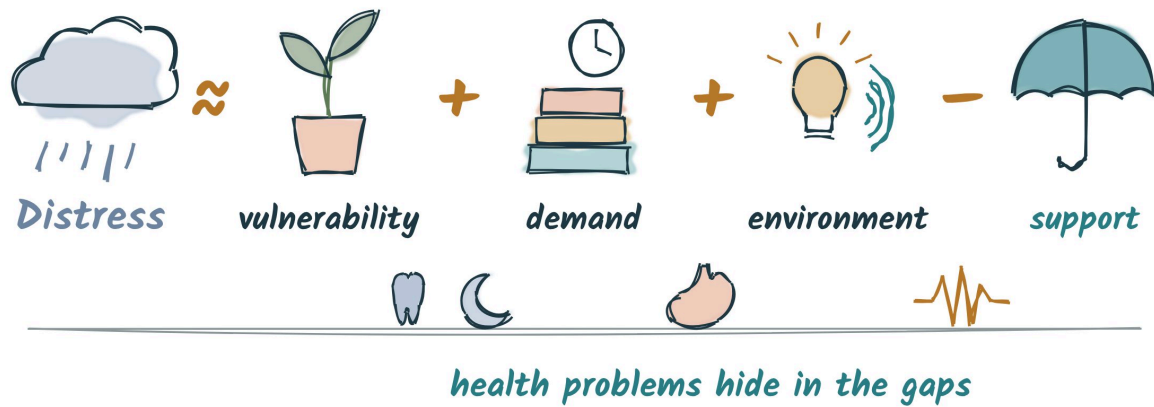
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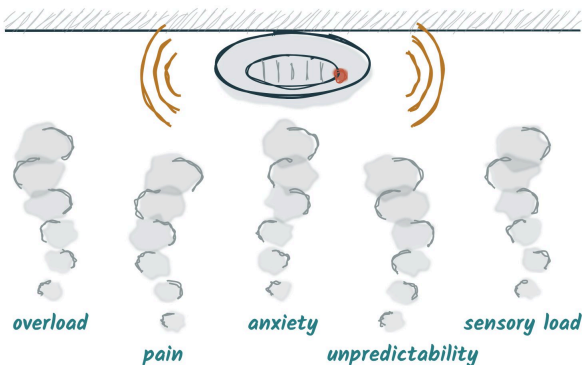
Closing (13:15–13:50) — From Genes to the School Gate: Understanding the Autistic Child in Daily Life

Four take-homes

- Behaviour is a clue, not the diagnosis.
- Distress ≈ vulnerability + demand + environment – support (health problems hide in the gaps).
- Do not diagnostic-overshadow: pain, sleep, constipation, epilepsy, anxiety, ADHD, ARFID still count.
- Goal ≠ looking less autistic — goal = function, communication, safety, belonging.



Behaviour · Clinic/A&E; adjustments · School distress



Behaviour is a clue, not the diagnosis.
ask why now, why here

- Meltdown/shutdown/refusal may signal overload, pain, anxiety, unpredictability, sensory load — ask why now, why here.
- Adjustments: first/last slot; quiet wait; one voice; explain-then-do; headphones/comfort object; parent as baseline interpreter; no forced eye contact; visual plan.
- Prefer “school distress” over “refusal”: sensory, social, masking crash, transitions, learning mismatch, sleep, pain, demand load, burnout.
- Validate; screen mood/risk/sleep/pain; graded school plan; avoid “just make them go”; involve EP/CAMHS/paeds/social care when needed.

Don't-miss (overshadowing)



Downloads & further reading

This handout and slides (when released): <https://practical-autism-research.co.uk> → Resources → CHER 2026.
 Genetics content and related materials on the same site. Book: *The Genetics of Autism* (Dr Mark Aszkenasy).