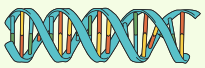
Beyond the Autism Diagnosis

Could your child's autism diagnosis really be a rare disorder? Getting the right diagnosis is important for proper treatment.

🔎 When Should We Look Closer?



Genetic clues



Unexpected Development



Atypical Features

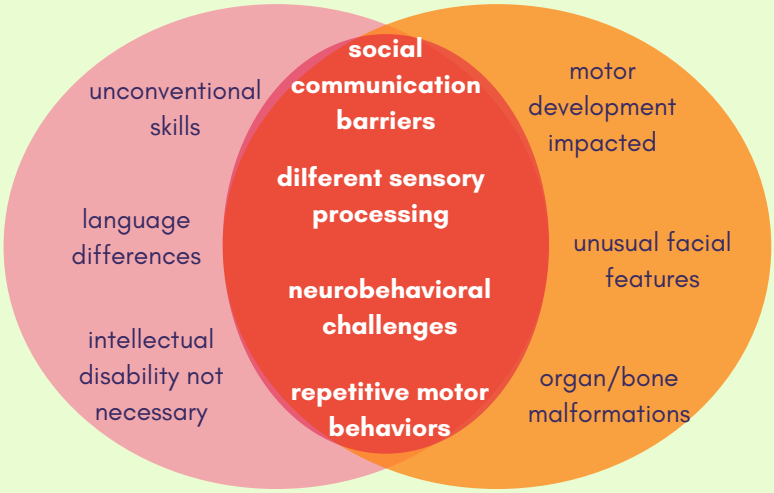


Social Difficulties



Intellectual Disability

Autism or Rare Disorder?



How is it Investigated?

Evaluation and Testing
Developmental and family history, genetic or other testing, and the overall clinical picture.

Specialist Input
Genetics, neurology, developmental pediatrics, or other relevant specialists.

Interpretation and Follow Up
Results are interpreted in context and guide diagnosis, management, and referrals.

If your child has any of the symptoms commonly found in a genetic disorder, consider testing.
Medicaid and private insurance cover comprehensive genetic testing when unexplained symptoms are present.

Some Examples Include...

Lujan-Fryns Syndrome



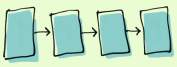
X-linked recessive
- Intellectual disability
- Marfanoid Habitus
- Nasal speech and hypotonia

Fragile X Syndrome



X-linked dominant
- Speech and language delay
- Intellectual disability
- Large ears and social anxiety

Rett Syndrome



X-linked dominant
- Often spontaneous
- Primarily affects girls
- Development advances then regresses

Cornelia de Lange Syndrome



Dominant
- Caused by many different genes
- Unusual facial features
- Growth and limb reduction

Resources

- upf3b.org
- UT Health San Antonio, Division of Pediatric Genetics
- Simons Searchlight
- NORD (National Organization of Rare Diseases)
- NCBI Gene Reviews
- Chromosome 18 Clinical Research Center, UT Health San Antonio

Developed by the UPF3B Project • Reviewed by Shayanne Martin MPH UT Health San Antonio • September 2026