SHANK3 Genetic Testing Program

GeneDx and Jaguar Gene Therapy have partnered to provide eligible pediatric and adult patients with suspected SHANK3-related autism spectrum disorder access to genetic testing. Through this program, denial of an insurance claim or lack of insurance coverage does not impact eligible patients' ability to access recommended testing.

Why participate?

Support for uncovered testing costs for eligible patients

Access to comprehensive exome sequencing from the leader in rare disease genomics

Clear, clinically actionable reports to guide care and next steps

Is your patient eligible? Patients must meet all of the following criteria:

- ☑ Have not had prior genetic testing performed by a clinical laboratory that resulted in a confirmed diagnosis of Phelan-McDermid Syndrome; SHANK3 haploinsufficiency.
- ☑ Present with moderate-to-severe developmental delay, intellectual disability (ID), autism spectrum disorder (ASD), or autistic-like behavior with clinical suspicion of PMS.
- ☑ Patient must reside in the United States.

Additionally, patients must meet at least 5 criteria from at least 2 of the below groups:

Neurology/neuropsychiatric:

- ☐ Psychiatric manifestations or episodes
- ☐ Seizures
- ☐ Regression
- ☐ Sleep disturbances
- ☐ Catatonia

Language/communication:

- ☐ Delayed or Absent Speech
- ☐ Speech apraxia

Sensory/sensory perception:

- ☐ Decreased perception of pain (including self-injury)
- ☐ Decreased response to auditory or visual stimuli, decreased perspiration/overheating, pica (and/or mouthing, chewing, or teeth grinding).

Dysmorphic features/musculoskeletal:

- ☐ Dysplastic fingernails or toenails, long eyelashes, large or fleshy hands
- ☐ Marked hypotonia

Motor:

- ☐ Delayed motor milestones (rolling over, sitting, crawling, walking)
- ☐ Gross and fine motor impairments
- ☐ Gait abnormalities

Gi/urinary system dysfunction:

- ☐ Bladder or bowel incontinence Gastroesophageal reflux (including difficulty swallowing)
- ☐ Dysmotility (including constipation)

How to order

1. Login or sign up for a GeneDx account and add 1 of the following ExomeDx™ tests to your cart.
   - ExomeDx, proband
   - ExomeDx, duo
   - ExomeDx, trio
2. Select the partnership code ESAS3
3. Confirm your patients' eligibility
4. Follow the prompts and enter in the appropriate information
5. Place your order and we'll follow up when your results are ready.

Please note: if exome results are non-diagnostic GeneDx will reach out to offer genome testing completely covered by Jaguar Gene Therapy

Clinicians can also order via a paper program TRF

For access to the program-specific TRF, please scan the QR code or visit genedx.com/partnership-programs/shank3-genetic-testing

A first-line test for autism

Exome sequencing is recommended as a first-line genetic test for individuals with autism spectrum disorder (ASD) when co-occurring features such as developmental delay, intellectual disability, or epilepsy are present, according to leading clinical guidelines.[1-3] GeneDx performs sequencing at a minimum of 10x coverage with an average depth of 100–120x across the exome. All samples are processed in GeneDx's CLIA-certified and CAP-accredited laboratory. For full test details, visit the GeneDx Test Catalog.

Remove barriers to testing. Empower earlier insights.

References:
1. Manickam K, McClain MR, Demmer LA, et al. Exome and genome sequencing for pediatric patients with congenital anomalies or intellectual disability: an evidence-based clinical guideline of the American College of Medical Genetics and Genomics (ACMG). Genet Med. 2021 Nov;23(11):2029-2037. doi: 10.1038/s41436-021-01242-6. Epub 2021 Jul 1.
2. Rodan LH, Stoler J, Chen E, et al. Genetic Evaluation of the Child With Intellectual Disability or Global Developmental Delay: Clinical Report. Pediatrics. 2025 Jun 23:e2025072219. doi: 10.1542/peds.2025-072219.
3. Smith L, Malinowski J, Ceulemans S, et al. Genetic testing and counseling for the unexplained epilepsies: An evidence-based practice guideline of the National Society of Genetic Counselors. J Genet Couns. 2022 Oct 24. doi.org/10.1002/jgc4.1646

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