

Autism Answers: *SHANK3* Inform Partnership 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, eligible patients can access recommended genetic testing even if they lack insurance coverage or their insurance claim is denied.



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 Autism Spectrum Disorder (ASD), standard cognitive score <50, and non-verbal or minimally verbal*, with clinical suspicion of PMS (**no words or primarily single words with occasional phrases*)
- ✓ Patient must reside in the United States
- ✓ Must have an ordering provider who is authorized under applicable law to order genetic testing in the United States
- ✓ Must have consented to the mandatory data sharing practices stated in the TRF

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

Neurology/neuropsychiatric

- Psychiatric manifestations or episodes (including bipolar disorder, schizophrenia / schizoaffective disorder)
- Seizures
- Regression
- Sleep disturbances
- Catatonia

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 ExomeDx™ proband to your cart
- 2 Select the partnership code ESAS3
- 3 Confirm your patient's 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.



Scan the QR code to get started ordering

For access to the program-specific TRF, please scan the QR code or visit genedx.co/shank3

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 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. 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.

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