JAGUAR GENE THERAPY | GeneDx

Introducing the

Autism Answers: *SHANK3* Inform Partnership Program

Providing access to exome and genome sequencing to eligible patients

Jaguar Gene Therapy and GeneDx have partnered to provide eligible patients with suspected undiagnosed Phelan-McDermid syndrome access to exome and genome sequencing.

The *SHANK3* Inform Partnership Program is designed to reduce barriers for accessing clinical guideline-recommended next-generation genetic testing to support potential earlier diagnosis of Phelan-McDermid syndrome in eligible patients. The program was designed in consultation with clinicians, researchers, and patient advocacy groups involved in the diagnosis and management of patients with Phelan-McDermid syndrome.

Why Genetic Testing?

Exome and genome sequencing are recommended as a first-line genetic test for pediatric patients with developmental delay, intellectual disability, and congenital anomalies, according to current clinical guidelines.[1,2] Older methods like chromosomal microarray (CMA) can miss smaller deletions or sequence variants in *SHANK3* leading to missed or false-negative diagnoses.[3]

About Phelan-McDermid syndrome[4-10]

Phelan-McDermid syndrome is the clinical diagnosis for *SHANK3* haploinsufficiency, which is caused by pathogenic variants in *SHANK3* or by chromosome 22q13.3 deletions encompassing the *SHANK3* gene. Individuals with Phelan-McDermid syndrome present with lifelong and severe neurobehavioral, communicative, motor, cognitive and social impairments characterized by delayed or absent developmental milestones. *SHANK3* haploinsufficiency is also a leading monogenic cause of autism spectrum disorder (ASD).

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Why participate?

- Support for genetic testing costs not covered by insurance for eligible patients
- Access to comprehensive exome sequencing
- Access to proband genome testing if proband exome results are non-diagnostic
- Clear, clinically actionable reports to guide care and next steps
- Post-test genetic counseling services offered through GeneDx at no cost

The Phelan-McDermid Syndrome Foundation and CureSHANK are dedicated to supporting those with Phelan-McDermid syndrome and their families.

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Currently, there are no FDA-approved treatments for Phelan-McDermid syndrome. Consensus guidelines on managing patients with Phelan-McDermid syndrome highlight recommendations for monitoring disease, symptom management that may include medication, behavioral intervention, speech and language therapy, physical therapy, and educational support.[8]

Remove barriers to testing. Empower earlier insights.

Phelan-McDermid syndrome is underdiagnosed due to challenges in current genetic testing practices, access limitations to genetic testing due to insurance coverage and cost, and gaps in clinical guidelines. Although professional society clinical guidelines recommend genetic testing for pediatric patients who present with key features of Phelan-McDermid syndrome such as developmental delay, intellectual disability, and ASD, some guidelines do not prioritize the most sensitive technologies, such as exome and genome sequencing.[1,2,8,9,10,11]

Is your patient eligible?

Eligibility criteria include:

1. No prior genetic testing performed by a clinical laboratory that resulted in a confirmed diagnosis of Phelan-McDermid syndrome (genetically confirmed *SHANK3* haploinsufficiency)
2. Patient must present with moderate to severe developmental delay, intellectual disability, ASD, or autistic-like behavior with clinical suspicion of Phelan-McDermid syndrome
3. Patient must reside in the United States

How to order?

- Visit the program page on GeneDx's website for more information on how to order genetic testing through the *SHANK3* Inform Partnership Program
- Testing can be ordered via the online portal or by downloading a program-specific paper Test Requisition Form at the link above
- For full test details, visit the GeneDx Test Catalog
- Additional resources are available at your request
  - Contact: medinfo@jaguargenetherapy.com

Complete eligibility criteria can be found on the program page on GeneDx's website or by using the following QR code.

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