JAGUAR GENE THERAPY | GeneDx

Introducing the

Autism Answers: *SHANK3* Inform Partnership Program

Providing access to exome sequencing for eligible patients

Jaguar Gene Therapy and GeneDx have partnered to provide eligible patients suspected of having Phelan-McDermid syndrome access to exome 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] Methods like chromosomal microarray (CMA) may not detect smaller deletions or sequence variants in *SHANK3* that may be detected by sequencing-based approaches.[3]

About Phelan-McDermid syndrome[4-10]

Phelan-McDermid syndrome is a rare neurodevelopmental disorder caused by *SHANK3* haploinsufficiency resulting from pathogenic variants in *SHANK3* or by chromosome 22q13.3 deletions that encompass *SHANK3*. 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).



Phelan-McDermid Syndrome Foundation

Why participate?

- Support for genetic testing costs not covered by insurance for eligible patients
- Access to exome sequencing
- Clinical genetic testing reports to inform 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 may be underdiagnosed due to due to barriers in being offered genetic testing, insurance coverage and cost of testing, and access to next-generation sequencing.[11] 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, recommendations regarding the type of genetic testing vary by guideline.[1,2,8,9,10,12]

Is your patient eligible?

Key eligibility criteria include:

1. No prior genetic testing performed by a clinical laboratory that resulted in a confirmed diagnosis of Phelan-McDermid syndrome (*SHANK3* haploinsufficiency)
2. Patient must present with all of the following: autism spectrum disorder (ASD), standard cognitive score <50, and nonverbal or minimally verbal*, 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

* no words or primarily single words with occasional phrases

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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