Additional DNA samples can help find answers

Your doctor has ordered comprehensive genetic testing (called exome or genome sequencing) and requested a sample from biological relatives.

Why are more samples needed?

Everyone's DNA is unique and contains natural variation. Comparing the patient's DNA with their biological relatives' DNA helps experts better understand which genetic findings may be important.

Including biological relatives' samples may help:

- Increase the chance of finding an answer for your child[1-4]
- Reduce uncertain results[5]
- Prevent delays from follow-up testing
- Help guide care sooner

With most insurance plans, biological relative samples are processed as part of one's genetic test rather than billed as separate tests.

Don't wait!

If you have been asked to provide a sample, please complete and return it as soon as possible. The sooner samples are received, the sooner testing can move forward.

References: 1. Clark MM, Stark Z, Farnaes L, et al. Meta-analysis of the diagnostic and clinical utility of genome and exome sequencing and chromosomal microarray in children with suspected genetic diseases. NPJ Genom Med. 2018 Jul 9;3:16. doi: 10.1038/s41525-018-0053-8. eCollection 2018. **2.** Lee H, Deignan JL, Dorrani N, et al. Clinical exome sequencing for genetic identification of rare Mendelian disorders. JAMA. 2014 Nov 12;312(18):1880-7. doi: 10.1001/jama.2014.14604. **3.** Farwell KD, Shahmirzadi L, El-Khechen D, et al. Enhanced utility of family-centered diagnostic exome sequencing with inheritance model-based analysis: results from 500 unselected families with undiagnosed genetic conditions. Genet Med. 2015 Jul;17(7):578-86. doi: 10.1038/gim.2014.154. **4.** Sawyer SL, Hartley T, Dyment DA, et al. Utility of whole-exome sequencing for those near the end of the diagnostic odyssey: time to address gaps in care. Clin Genet. 2016 Mar;89(3):275-84. doi: 10.1111/cge.12654. **5.** Rehm H, Alaimo JT, Aradhya S, et al. The landscape of reported VUS in multi-gene panel and genomic testing: Time for a change. Genet Med. 2023 Jul 30;100947. doi: 10.1016/j.gim.2023.100947.

Las muestras adicionales de ADN pueden ayudar a encontrar *respuestas*

Su médico ha solicitado una prueba genética integral (llamada secuenciación del exoma o secuenciación del genoma) y ha pedido una muestra de familiares biológicos.

¿Por qué se necesitan más muestras?

El ADN de cada persona es único y presenta variaciones naturales. Comparar el ADN del paciente con el de sus familiares biológicos permite que los especialistas comprendan mejor qué hallazgos genéticos podrían ser importantes.

La inclusión de muestras de familiares biológicos tiene los siguientes beneficios:

- Aumentar las probabilidades de encontrar una respuesta para su hijo(a)[1-4].
- Reducir la cantidad de resultados inciertos[5].
- Evitar retrasos ocasionados por pruebas de seguimiento.
- Ayudar a orientar la atención médica más rápidamente.

En la mayoría de los planes de seguro médico, las muestras de los familiares biológicos se procesan como parte de la prueba genética del paciente y no se facturan como pruebas independientes.

¡No espere!

Si se le ha solicitado proporcionar una muestra, envíela lo antes posible. Cuanto antes se reciban las muestras, antes podrá iniciarse el análisis.

Referencias: 1. Clark MM, Stark Z, Farnaes L, et al. Meta-analysis of the diagnostic and clinical utility of genome and exome sequencing and chromosomal microarray in children with suspected genetic diseases. NPJ Genom Med. 9 de julio de 2018;3:16. doi: 10.1038/s41525-018-0053-8. eCollection 2018. **2.** Lee H, Deignan JL, Dorrani N, et al. Clinical exome sequencing for genetic identification of rare Mendelian disorders. JAMA. 12 de noviembre de 2014;312(18):1880-7. doi: 10.1001/jama.2014.14604. **3.** Farwell KD, Shahmirzadi L, El-Khechen D, et al. Enhanced utility of family-centered diagnostic exome sequencing with inheritance model-based analysis: results from 500 unselected families with undiagnosed genetic conditions. Genet Med. Julio de 2015;17(7):578-86. doi: 10.1038/gim.2014.154. **4.** Sawyer SL, Hartley T, Dyment DA, et al. Utility of whole-exome sequencing for those near the end of the diagnostic odyssey: time to address gaps in care. Clin Genet. Marzo de 2016;89(3):275-84. doi: 10.1111/cge.12654. **5.** Rehm H, Alaimo JT, Aradhya S, et al. The landscape of reported VUS in multi-gene panel and genomic testing: Time for a change. Genet Med. 30 de julio de 2023;100947. doi: 10.1016/j.gim.2023.100947.