



The Quinn Madeleine Foundation establishes first-ever Niemann-Pick diagnostic carrier screening program

Aiming to eradicate fatal ASMD, The Quinn Madeleine Foundation has partnered with Baby Genes to create a diagnostic carrier screening program for at-risk individuals.

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The Quinn Madeleine Foundation has partnered with Baby Genes Inc. to provide diagnostic carrier screening at no charge to hundreds of people impacted by Acid Sphingomyelinase Deficiency (ASMD), also known as Niemann-Pick Disease Types A & B, a severe lysosomal storage disorder. Type A (NPA) is always fatal by toddlerhood.

Baby Genes' gene sequencing technology provides accurate, efficient results identifying any disease-causing mutation, not just the common mutations recognized by traditional carrier screenings. Today there is neither treatment nor a cure for NPA. Providing access to this test will enable prospective parents to make the best, most informed decisions in their family planning. "Our unified goal is to facilitate the discovery of new molecular targets and provide actionable information to those bringing new therapy options to patients and families," said President/CEO of Baby Genes, Inc. Rich Sjogren.

The program is currently open to any family member of an affected child, though it intends to go further. "The long-term goal is to consider the possibility of an ethnicity pattern, identifying a higher-risk general population and offering more widespread screening," says Eileen Linzer, Co-Founder & Executive Director of The Quinn Madeleine Foundation. Clients screened are asked to complete a self-reported Ethnicity Survey to assist in this research.

To learn more, contact The Quinn Madeleine Foundation at genetics@quinnmadeleine.org.

About The Quinn Madeleine Foundation

The Quinn Madeleine Foundation is a recognized 501(c)(3) non-profit charity. Wholly volunteer run, QMF seeks to aid in the prevention of Niemann-Pick Disease Type A, a rare genetic disease that is always fatal in toddlerhood. The Quinn Madeleine Foundation also serves as a wish-granting organization for children under age 3 who have been diagnosed with a terminal or life-threatening disorder. The Quinn Madeleine Foundation is located in Lynbrook, NY and their website is www.quinnmadeleine.org.

About Baby Genes

Baby Genes Inc is a CLIA-certified, diagnostic laboratory that offers advanced newborn testing using Next-Generation Sequencing (NGS). The Baby Genes Newborn Panel tests 106 genes for abnormalities that are linked to 171 hereditary disorders. The Baby Genes Newborn Test can be used as a confirmatory/reflex test for kids with abnormal NBS results, as a supplement to state-based newborn screening or as a family planning/carrier test. Baby Genes is located in Golden, CO and their website is www.babygenes.net.