

Écrit par CENTOGENE

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Collaboration between FDNA, the developer of the Face2Gene Suite and CENTOGENE, worldwide leader in genetic diagnostics, enabled the first grant of molecular testing for a patient with an unknown genetic disorder. The collaboration extends beyond financial support and includes the successful integration of Face2Gene LABS API with CENTOGENE's bioinformatics pipeline to improve variant prioritization and analysis of patients with undiagnosed genetic disorders using deep phenotyping and facial analysis technology.

FDNA Inc., developer of the Face2Gene suite of phenotyping apps, announced today the successful results of its grant program to use the Face2Gene deep phenotyping and facial analysis technology to support the variant prioritization and analysis of molecular testing of patients with unknown genetic disorders. This announcement was made at the American Society of Human Genetics (ASHG) 2016 in Vancouver, Canada. Through this grant program, CENTOGENE, a worldwide leader in the field of genetic diagnostics for rare hereditary diseases, became the first commercial lab to join FDNA by sponsoring molecular testing for patients with unknown genetic disorders.

After launching the grant program in March 2016, the case was uploaded to Face2Gene's Unknown Forum and reviewed by an expert review panel (ERP) made up of clinical geneticists that review and share insights and expertise on undiagnosed and challenging cases. The case was identified by expert geneticists from the ERP as a qualified candidate for the grant based on various criteria such as patient's access to genetic testing and potential medical benefits from exome sequencing. FDNA and CENTOGENE collaborated to offer molecular testing for the patient free of charge. This is the first genetic testing grant awarded to a patient through FDNA's collaborative grant program.

"We are proud to collaborate with one of the world's best laboratories in our effort to bring hope to patients and help enable successful diagnoses of rare and ultra-rare genetic disorders," said Dekel Gelbman, FDNA's Chief Executive Officer. "Typically, this process can cost thousands of dollars and, for many families, isn't accessible. Our grant program's mission will continue to bring a positive impact in the lives of patients living undiagnosed with rare and ultra-rare genetic diseases."

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“Unidentified diseases can create a significant burden on patients and their families, especially when the means to properly diagnose the disease are not within their financial reach,” said Prof. Peter Bauer, COO of CENTOGENE. “It’s an honor for us to join forces with FDNA to help provide these patients with meaningful answers and help to improve their quality of life, while paving the road for proper disease management.”

Individuals and institutions interested in learning more about these grants, or who wish to partner with FDNA in this initiative can contact FDNA at info@FDNA.com, or stop by FDNA booth #1309/CENTOGENE booth #1317 at the American Society of Human Genetics (ASHG) Annual Meeting in Vancouver, October 18-22.

About Face2Gene

Face2Gene is a suite of phenotyping apps that facilitates comprehensive and precise genetic evaluations, including: CLINIC - a utility that enhances patient evaluation with deep phenotyping; FORUMS – a platform for collaborative case review for diagnostic dilemmas; LIBRARY | LONDON MEDICAL DATABASES – a resource for trusted dysmorphology content; LABS – an API that enables better variant analysis through deep phenotyping; and ACADEMY – an interactive dysmorphology training tool.

For more information, please visit <http://www.Face2Gene.com>

About FDNA

FDNA’s mission is to save lives and improve the quality of life of patients with rare or difficult-to-diagnose genetic syndromes. FDNA has developed the Face2Gene suite of phenotyping apps that facilitate comprehensive and precise genetic evaluations.

For more information, please visit <http://www.FDNA.com>

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