



USP7 Foundation Collaborates with FDNA to Solve Medical Mysteries

September 12, 2018 – FALMOUTH, ME – The Foundation for USP7 Related Diseases, in collaboration with [FDNA](#), announces the successful recognition of the facial phenotype of USP7 syndrome using Face2Gene, FDNA’s suite of phenotyping applications. This cooperative effort between the USP7 Foundation and FDNA is part of the Genomics Collaborative, an FDNA initiative to accelerate breakthroughs in precision medicine using deep learning and artificial intelligence.

With less than 40 known cases around the world, USP7 related disease arises from mutations of USP7, a protein-coding gene that plays a role in tumor suppression, transcriptional regulation, immune response, and endosomal protein recycling. Symptoms include autism, epilepsy, speech impairment, intellectual disability, and significant feeding problems, among others. The ability to identify the facial phenotype of USP7 earlier in development enables more precise and personalized treatments and leads to improved quality of life.

“Because there is such a small population of USP7 patients, my own daughter included, having a tool such as Face2Gene trained to recognize this syndrome increases awareness and likelihood of an earlier diagnosis,” said Bo Bigelow, Chairman and Co-founder of the USP7 Foundation. “We’re excited to see the results of this research and are looking forward to continuing our work with the Genomics Collaborative and expanding our community to improve treatment for those living with this disease.”

FDNA’s next-generation phenotyping (NGP) technologies that capture, structure, and interpret complex physiological information, are used in analyzing patient clinical data and next-generation sequencing (NGS) data. The outcome is real-time discovery of disease biomarkers, advancement of clinical and molecular technologies, and the creation of effective and personal treatments.

“We’re thrilled by the outcome of this successful integration with our technology,” said Ilana Jacqueline, Patient Advocacy Manager at FDNA and Coordinator of the Genomics Collaborative. “Through the use of artificial intelligence and collaborations such as this, we’re one step closer to eliminating diagnostic delay.”

The Foundation for USP7 Related Diseases seeks to cure those diagnosed with a mutation of USP7. They do this in two ways: funding research efforts, and identifying and increasing our work with patients living with the disease.

[Learn more about the Foundation for USP7 Related Diseases and ways to get involved.](#)

The Foundation for USP7 Related Diseases was founded in 2017. To further its mission, it raised nearly \$100,000 for research in its first year, which will fund a phenotyping study at Baylor College of Medicine in Houston, TX. The foundation’s other current effort, entitled Project Artemis, aims to identify 100 USP7 patients worldwide. “We’re optimistic that Face2Gene will reach undiagnosed families, add to our patient count, and help us reach our goal in the coming year,” said Bigelow.