

NextGeneration Phenotyping: A Performance Analysis of this Innovative Research Platform

In their latest study being presented at the National Organization for Rare Disorders, FDNA's Face2Gene software has shown over 98% success rate in matching 100 of the most prevalent rare diseases with an accurate diagnosis. Face2Gene is revolutionizing the accessibility and aggregation of data for the more than 7,000 rare diseases impacting 350 million people globally with their use of next-generation phenotyping, including facial analysis and artificial intelligence.

After training Face2Gene's artificial intelligence to recognize the visual markers of various rare diseases and genetic syndromes with tens of thousands of patient images, each image was analyzed and ranked according to similarities with identified 'syndrome classifiers.' To measure success, FDNA used the area under the curve (AUC) of the receiver operating characteristic (ROC) curve. With an AUC of 100% representing perfect accuracy, FDNA assessed the accuracy of all syndrome classifiers in the Face2Gene system.

FDNA's results of near-perfect performance among hundreds of syndrome classifiers prove the valuable utility of Face2Gene for clinicians, genetic counselors and patients in decreasing the time and cost of diagnosis of rare diseases.

For more information on FDNA's work towards eliminating the diagnostic journey of patients with rare diseases, or to set up an interview with CEO Dekel Gelbman, please contact fbasile@cerconebrown.com