Instead of a press release summarizing the findings of poster

Next-Generation Phenotyping (NPG) for Cat-Eye Syndrome

Here the requested 3-4 paragraphs summarizing the research findings

Many genetic syndromes are associated with the appearance or gradual development of distinctive facial characteristics in affected patients. One of these syndromes is the so-called cat-eye-syndrome, which is (among others) characterized by iris coloboma, preauricular pits, and anal atresia. Besides, in cytogenetic analyses one can find a normal chromosome content of 46 plus a 47^{th} small supernumerary marker chromosome. This results in the presence of 4 instead of 2 copies of a specific genetic region derived from chromosome 22, and this gain of copy numbers is considered to be the reason for the clinical findings in cat-eye-syndrome.

With around 250 reported cases cat-eye-syndrome is for sure under-reported as well as underdiagnosed. Nonetheless, this disease belongs for sure to the group of rare genetic disorders, or Orphan-diseases. Typical for carriers of such rare genetic conditions is normally a long odyssey from recognizing that ‘something may be wrong with my child’ until getting the correct diagnoses. However, only with a correct diagnosis latter adequate treatment options may be discussed with and offered to the patients and their families. The aim of the present project is to help families with an affected child known or not known to carry a small supernumerary marker chromosome, to find the diagnoses cat-eye-syndrome more quickly and more easily.

We take advantage of the FDNA technology used by thousands of clinical, research and lab sites globally in the clinical genomics space. More specifically, FDNA’s next-generation phenotyping (NGP) technology is used, which can capture, structure, and analyze complex human physiological data to produce actionable genomic insights. In other words, in this project we ask families to provide frontal facial pictures of cat-eye-syndrome patients, to be unloaded completely anonymously and in a way that providers can decide exactly if at all and who is allowed to see original pictures. Based on these well-defined pictures (provided together with the age of the corresponding patient when the picture was made) FDNA’s flagship product, Face2Gene - DeepGestalt module can in the end analyze facial photos of patients to highlight phenotypic similarity to known, but rare genetic disorders. It analyzes facial photos of patients to produce a list of syndromes that are found to have similar facial characteristics with accuracies of above 90%. DeepGestalt currently supports already more

than 300 genetic syndromes and syndromes groups, representing 45% of cases solved by whole exome sequencing. With this project one more syndrome, i.e. cat-eye-syndrome shall be added.

What do you have to do to help? You are invited to upload or send in facial photos of patients with confirmed diagnosis of genetic syndromes to a secure and private portal dedicated to help training the technology under https://fs27.formsite.com/Face2Gene/fvshgv9vv0/index.html. Serial photos of the same patient at different ages are particularly useful for showing changes over time, so you are encouraged to provide current and past photos. For each photo, please write the estimated age at the time the photo was taken, gender and ethnicity as well as the exact confirmed diagnosis (name of syndrome, specific test results, if available). This call is essential part of the project: “Establishment of next generation phenotyping in cat eye syndrome patients” supported by NORD Research Program (18002); responsible scientist. Prof. Thomas Liehr, Jena, Germany; Thomas.Liehr@med.uni-jena.de .