3D facial phenotyping in rare diseases

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Abstract :

Facial malformations are a frequent component of rare diseases, often associated with other organ disorders. From a clinical point of view, these dysmorphies can be in the foreground, responsible for symptoms of the orofacial sphere. They can also be discreet and go unnoticed until the syndromic diagnosis, on the occurrence of an extra-facial complication. The study of facial malformations thus remains a diagnostic challenge in certain rare diseases.

Based on a rare disease model, Stickler syndrome, a collagenopathy mainly responsible for ophthalmological complications, we propose a method for the description and analysis of pathological facial phenotypes using innovative and still little used tools in this indication: 3D camera and geometric morphometry.

This study involves two prospective cohorts: a control population (N=50) and a population of patients with Stickler syndrome (N=30).

We provide a description of the analysis process and an overview of the initial results. The analyses are still in progress at the time of submission of this abstract.