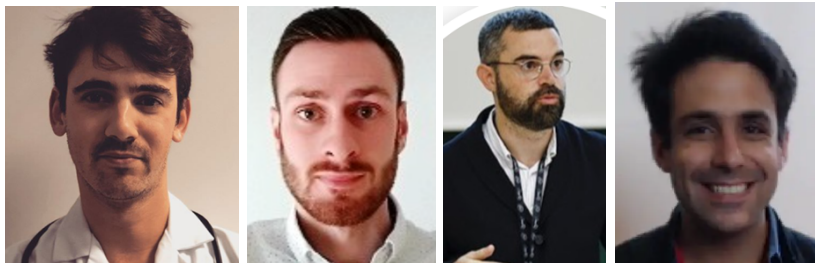




## AI-based diagnosis and phenotype understanding in Craniofacial Microsomia (CFM) using 2D photographs

### Authors :

Quentin Hennocq<sup>a,b</sup>, Thomas Bongibault<sup>a</sup>, Georges Carpentier<sup>c</sup>, Pierre Corre<sup>d</sup>, François Djate Delbrah<sup>a</sup>, Maxime Douillet<sup>a</sup>, Eva Galliani<sup>b</sup>, Wuttichart Kamolvisit<sup>e,f</sup>, Stanislas Lyonnet<sup>a,g</sup>, Thantrira Porntaveetus<sup>e,f</sup>, Sandrine Touzet-Roumazeille<sup>c</sup>, Arnaud Picard<sup>b</sup>, Marlène Rio<sup>a,g</sup>, Nicolas Garcelon<sup>a</sup>, Roman H. Khonsari<sup>a,b</sup>



### Institutions :

a. Imagine Institute, INSERM UMR1163, Paris, France

b. Département de chirurgie maxillo-faciale et chirurgie plastique, Hôpital Necker – Enfants Malades, Assistance Publique – Hôpitaux de Paris ; Centre de Référence des Malformations Rares de la Face et de la Cavité Buccale MAFACE, Filière Maladies Rares TêteCou ; Faculté de Médecine, Université de Paris Cité ; Paris, France.

c. University Lille, CHU Lille, Inserm, Service de Chirurgie Maxillo-Faciale et Stomatologie, U1008-Controlled Drug Delivery Systems and Biomaterial, Lille, France.

- d. INSERM U1229 - Regenerative Medicine and Skeleton RMeS, Nantes, France; Nantes University, CHU Nantes, Department of Oral and Maxillofacial Surgery, Nantes, France.
- e. Center of Excellence for Medical Genomics, Department of Pediatrics, Faculty of Medicine, Chulalongkorn University, Bangkok, Thailand
- f. Excellence Center for Genomics and Precision Medicine, King Chulalongkorn Memorial Hospital, The Thai Red Cross Society, Bangkok, Thailand
- g. Fédération de médecine génomique, Hôpital Necker – Enfants Malades, Assistance Publique – Hôpitaux de Paris ; Faculté de Médecine, Université de Paris Cité ; Paris, France.

**Abstract :**

Two to three thousand syndromes include facial anomalies. Their detection requires the experienced eye of an expert in dysmorphology. Here we present an innovative tool dedicated to the screening of abnormalities in children faces developed using a unique clinical photographic database.

The process was based on the automatic detection of landmarks, superposition of point clouds, dimension reduction and application of different classification algorithms. Our objectives were: 1) diagnose syndromes, 2) understand phenotypic features involved in diagnosis, 3) establish phenotypic differences within the same syndrome.

Craniofacial Microsomia (CFM) was used as an illustration of this process. The model allowed to predict the existence of extra-facial abnormalities based on frontal and facial 2D photographs.

Further application is currently being developed, involving the external ear, the eyes, the tip of the nose, and hair implantation.