

## **Diagnostic tools for craniofacial malformations based on artificial intelligence : are morphometric and cephalometric approaches still relevant ?**

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Shape analysis on 2D pictures can involve computations based on pre-defined rules; in machine learning approaches, these rules are replaced by the ability of the model to learn from examples.

Examples of relevant inputs in healthcare are X-rays, texts, pathology slices and clinical photographs. Among these inputs, clinical photographs are particularly important in the diagnosis of craniofacial malformations. Many among these 2-3000 syndromes have minor facial features or are very uncommon: their screening requires the eye of an expert in dysmorphology.

Moreover, some syndromes group under the same entity different genetic origins, such as Treacher Collins (TC) syndrome, with mutations in the TCOF1 or POLR1D genes. In this work, we use machine learning methods to group patients in different severity clusters of TC syndrome, and to understand if these differences in severity can be explained by differences in genotype. The idea is to anticipate the severity of the malformation to better manage it.