

Stickler syndrome 3D facial phenotype: morphometric analyses and diagnostic prospects

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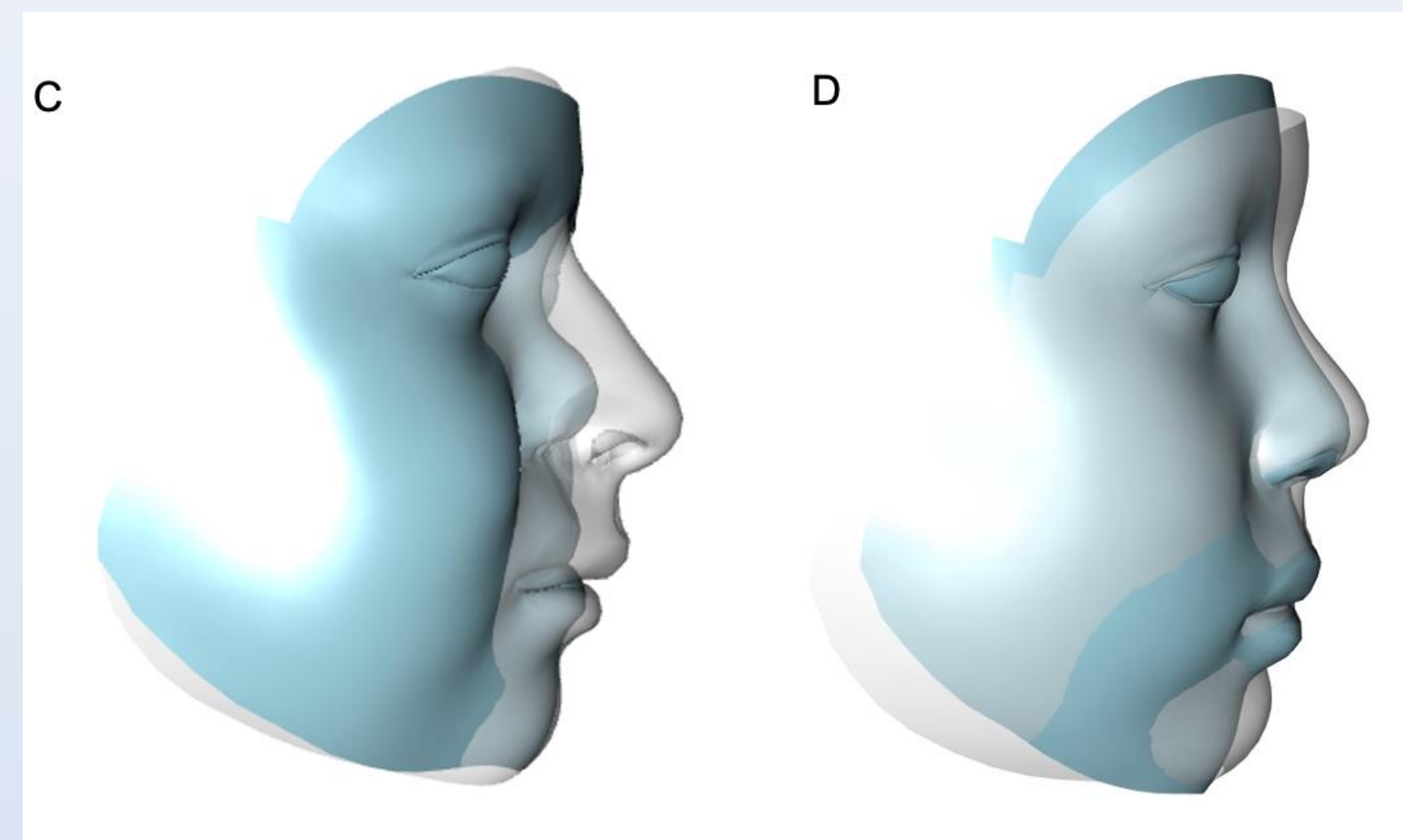
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Introduction

- Rare collagenopathy, incidence 1/7500
- Ubiquitous symptomatology with ophthalmological features predominance
- Significant diagnostic delay due to clinical polymorphism
- High incidence of ophthalmological complications (retinal detachment)
- Cause of visual impairment from early childhood
- Existence of a typical but subtle facial morphology in some Stickler patients, which could help diagnose them earlier



Stickler dysmorphism is characterised by proptosis associated with a significant nasal root depth and nasal underprojection

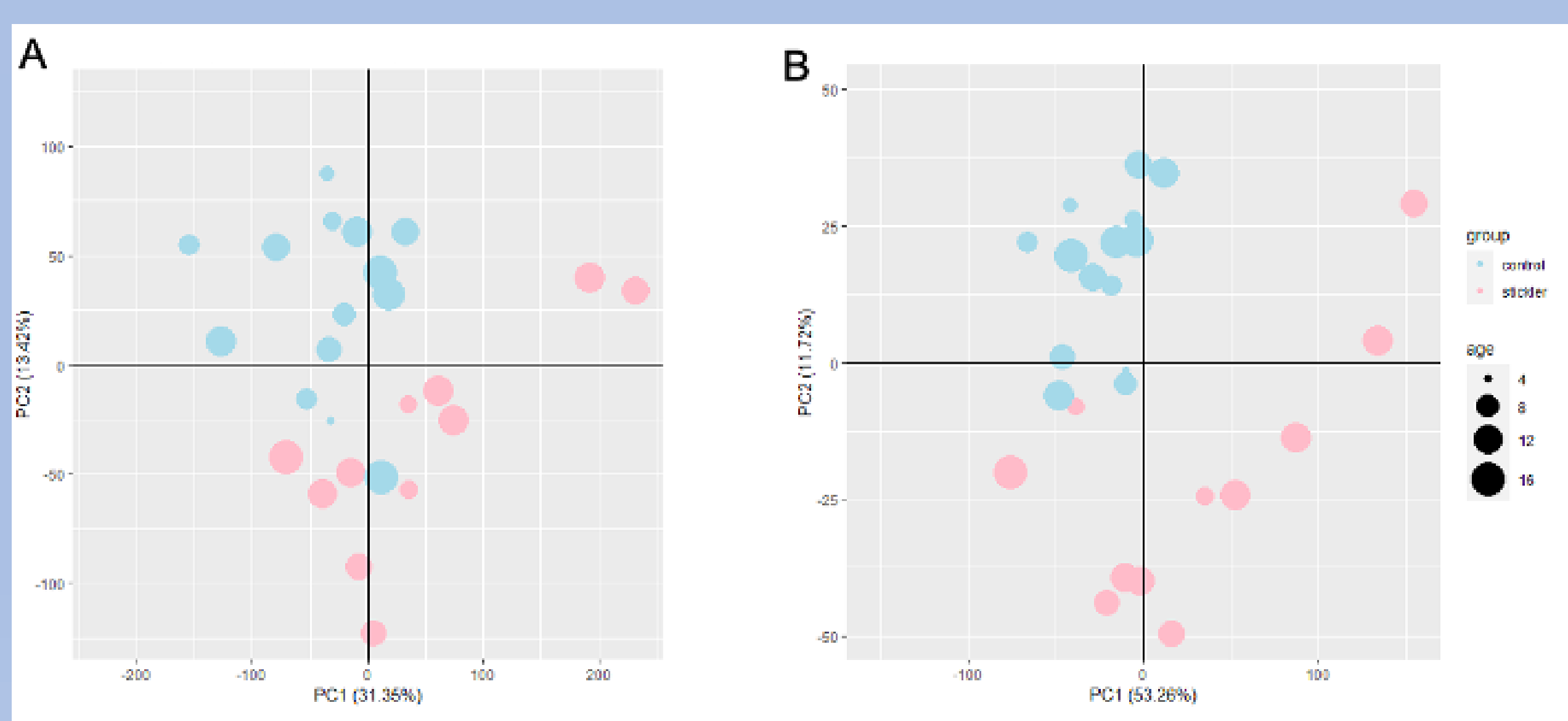
Objectives

To objectify Stickler syndrome dysmorphism
To describe its evolution over the growth
To assess an IA model in classify Stickler children from control children based on facial morphology

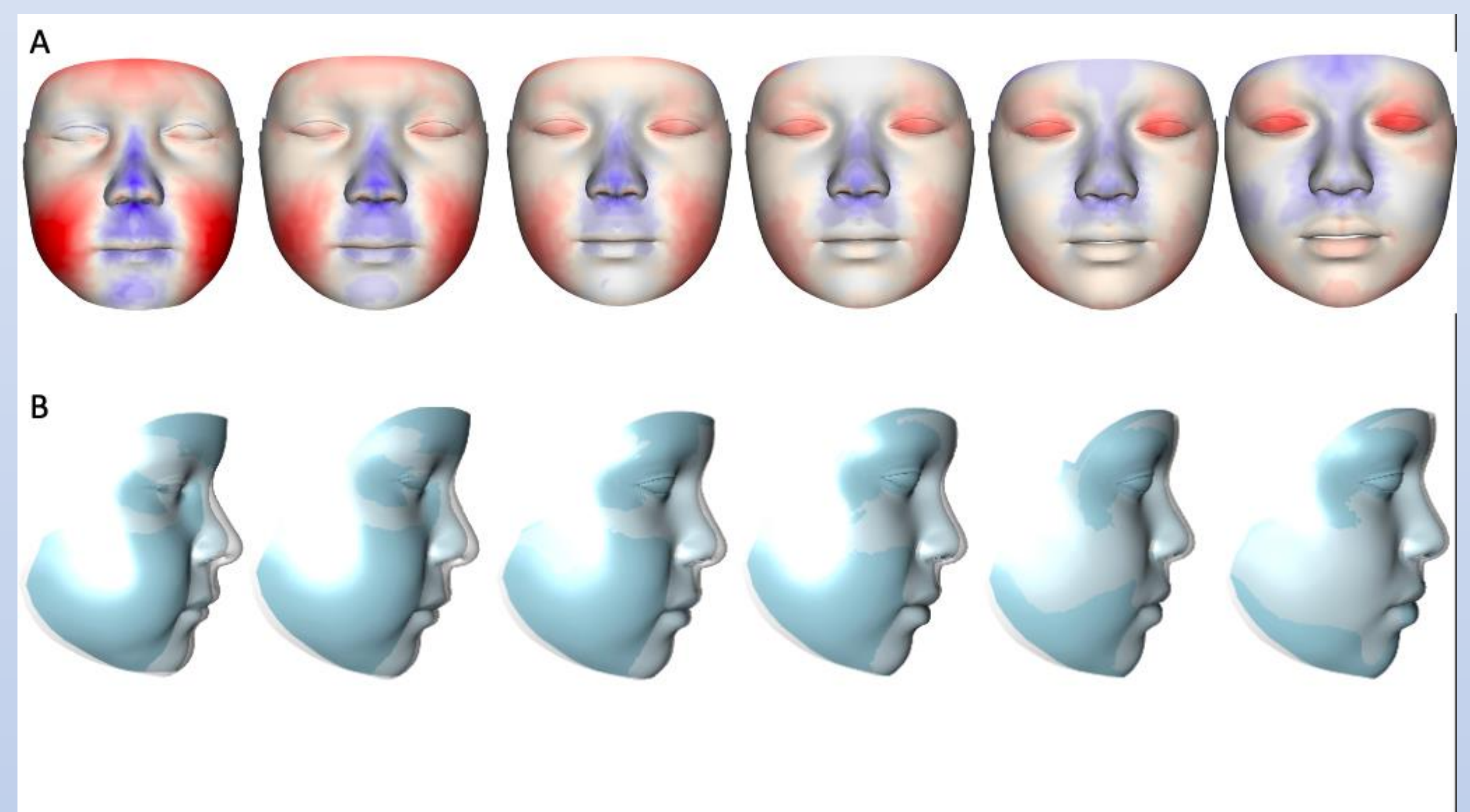
Methods

- Stickler children facial surfaces were collected by taking 3D photographs (11 patients) and control children by segmenting CT-scans (14 patients)
- Template-based registration of all facial surfaces was realized to standardize data
- Comparison was made using Principal Component Analyses (*R-software*)
- Comparative growthmodel between controls and Stickler children (*PLS2b – R software*)
- Support Vector Machine IA model was used to classify Stickler vs control based on facial morphology

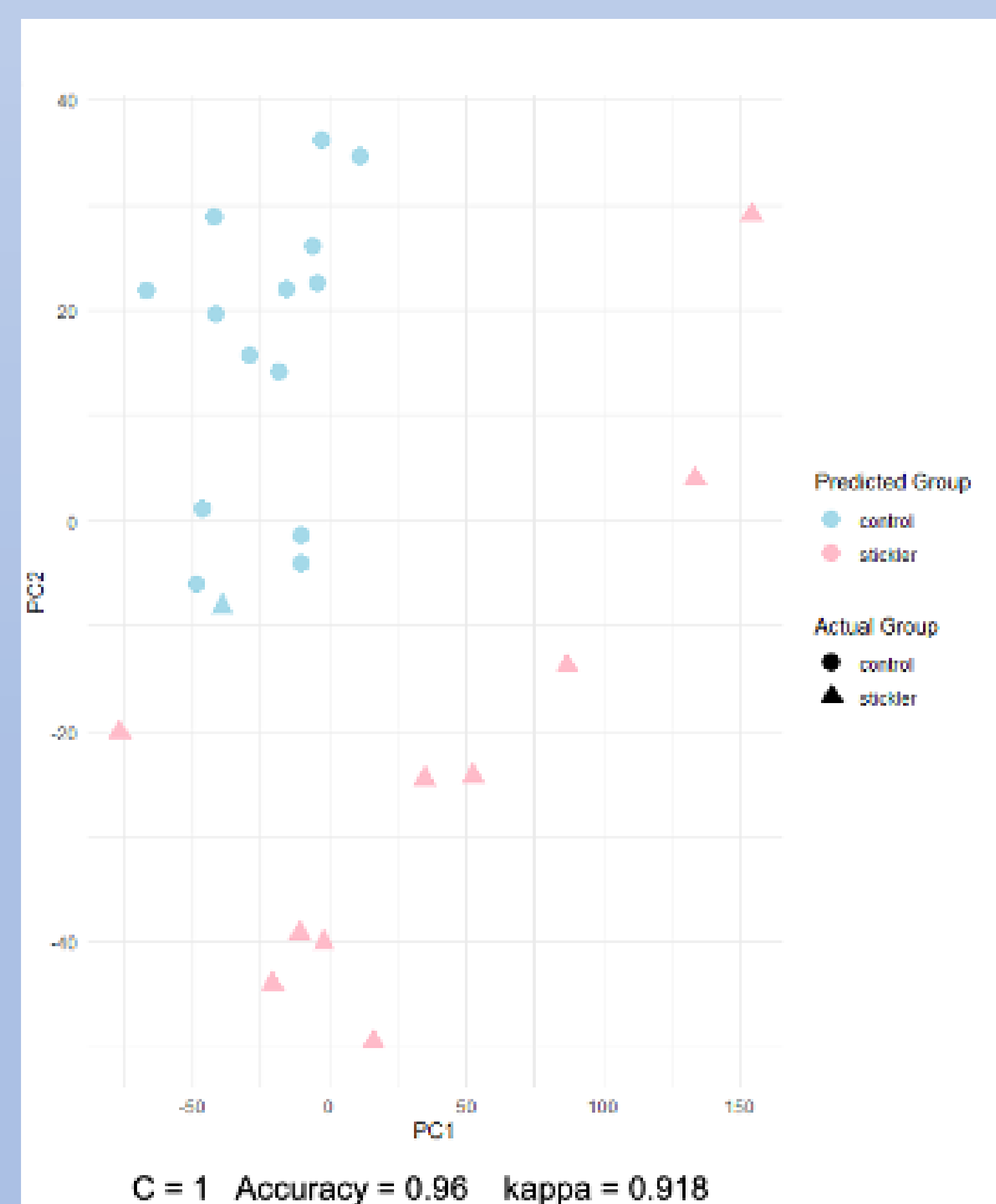
Results



PCA identified 2 populations corresponding to Stickler and control patients



Centrofacial hypoplasia lower with age while proptosis tends to increase. Stickler dysmorphism globally mitigates over time, to become difficult to identify



Only one Stickler patient was misclassified by SVM model.

Conclusion

Combining 3D photography with artificial intelligence has demonstrated its accuracy in identifying subtle phenotypic traits in syndromic patients. This can aid in the early detection of Stickler patients, enabling proactive measures to prevent ophthalmological complications.