# Abstract

The aim of the thesis was to evaluate facial dysmorphism in Williams (WBS), Noonan (NS) and DiGeorge syndrome (DGS) patients and also to evaluate changes in the morphology of the face during growth. In total 57 3D facial scans of patients of all ages were analysed, including 12 WBS, 20 NS, 25 DGS and 31 scans of control subjects. The evaluation has been carried out using methods of geometric morphometry, namely by coherent point drift - dense correspondence analysis, superprojection of mean faces, per vertex t-test and principal component analysis.

Statistically significant differences in the facial morphology were shown for all the syndromes vs. control. Observed dysmorphies in WBS (narrow forehead, bitemporal narrowing, periorbital fullness, bulbous and anteverted nasal tip, malar flattening, protrusion of both lips, pointed chin) mostly confirmed existing knowledge of the typical phenotype. The morphology in WBS is thus strongly specific and manifested in most of the patients. During ontogeny, the dysmorphic features associated with increased facial convexity become pronounced, while the other typical features remain relatively stable. In contrast to the control, the retrusion of the chin occurs during the development. Observed dysmorphic traits in NS (less prominent supraorbital ridges, periorbital fullness, small and broad nose with concave profile, depressed root and anteverted tip, narrow bizygomatic width, upper lip protrusion, micrognathia, deficient chin button) are mostly typical in childhood, then there was a noticeable reduction of their coarse appearance with increasing age. Facial development occurs in a similar way to the control; however, the area of the nose and chin increases more significantly by the adulthood. Facial characteristics observed in DGS, except for the hypoplastic alae nasi, do not correspond to the description of a typical facial appearance. This implies significant heterogeneity in their typical facial phenotype. Within the age changes, patients exhibit smaller effect of increasing the facial convexity, which is related to the more striking growth of their chin by adulthood.

It was demonstrated that the 3D methods used can be successfully applied in the study of dysmorphology.



**Keywords**: facial shape and size, surface model, ontogenetic trajectory, 3D imaging, geometric morphometry, dysmorphology, Williams syndrome, Noonan syndrome, DiGeorge syndrome