

# Dentofacial morphology in Turner syndrome karyotypes

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av

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leg. tandläkare

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Det medisinsk-odontologiske fakultet, Universitetet i Bergen, Bergen, Norge

Avhandlingen baseras på följande delarbeten:

- I. Rizell S, Barrenäs ML, Andlin-Sobocki A, Stecksén-Blicks C, Kjellberg H. 45,X/46,XX karyotype mitigates the aberrant craniofacial morphology in Turner syndrome. *European Journal of Orthodontics* 2012 Apr 24. (Epub ahead of print)
- II. Rizell S, Barrenäs ML, Andlin-Sobocki A, Stecksén-Blicks C, Kjellberg H. Palatal height and dental arch dimensions in Turner syndrome karyotypes. Submitted for publication
- III. Rizell S, Barrenäs ML, Andlin-Sobocki A, Stecksén-Blicks C, Kjellberg H. Turner syndrome isochromosome karyotype correlates with decreased dental crown width. *European Journal of Orthodontics* 2012 Apr;34(2): 213-8.
- IV. Rizell S, Kjellberg H, Dietz W, Norén JG, Lundgren T. Altered inorganic composition of dental enamel and dentin in primary teeth from girls with Turner syndrome. *European Journal of Oral Sciences* 2010 Apr;118(2): 183-90.



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# ABSTRACT

## Dentofacial morphology in Turner syndrome karyotypes

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The overall aim of this thesis was to study dentofacial morphology in Turner syndrome (TS) versus controls and the influence hereupon from karyotype.

One hundred thirty two TS females (5-66 years of age), from Göteborg, Uppsala and Umeå were participating. Cephalometric analysis, cast model analysis concerning palatal height, dental arch morphology and dental crown width were performed. Eighteen primary teeth were analysed in polarized light microscopy, scanning electron microscopy, microradiography and X-ray microanalysis were performed. The TS females were divided according to karyotype into: 1 45,X; 2 45,X/46,XX; 3 isochromosome; 4 other.

Compared to healthy females, TS were found to have a flattened cranial base as well as small and retrognathic jaws with a posterior inclination. The maxillary dentoalveolar arch was narrower and longer, while the mandibular dental arch was wider and longer in TS compared to controls. The palatal height did not differ comparing TS and healthy females. The dental crown width was smaller in TS for both permanent and primary teeth. Aberrant elemental composition, prism pattern and lower mineral density were found in TS primary enamel compared to enamel in primary teeth from healthy girls.

Turner syndrome karyotype was found having an impact on craniofacial morphology, with the mosaic 45,X/46,XX exhibiting a milder mandibular retrognathism as well as fewer cephalometric variables differing from controls compared to other karyotypes. Also for the dentoalveolar arch morphology the 45,X/46,XX group had fewer variables differing from healthy females. The isochromosome TS group exhibited the smallest dental crown width for several teeth, while 45,X/46,XX had the largest dental crown width for some teeth and fewer teeth than both 45,X and isochromosomes that differed from controls. Thus, the mosaic 45,X/46,XX seemed to exhibit a milder phenotype, possibly due to presence of healthy 46,XX cell lines.

**Keywords:** Orthodontics, genetics, Turner syndrome, karyotype, geno-phenotype correlation, anthropometrics, craniofacial morphology, dental arch, dental crown width, enamel, primary teeth, elemental composition

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