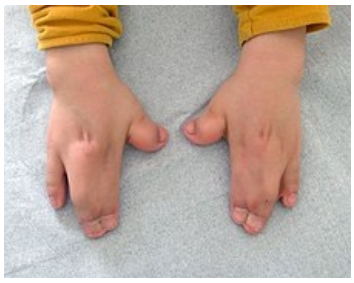
Apert syndrome

Apert's syndrome (*acrocephalosyndactyly*) is a rare genetic conditional disease affecting mainly the skull with premature **closure of the seams** and then the upper and lower limbs **syndactyly**. It belongs to the broad group of *craniosynostoses*, historically Apert syndrome and some other diseases from this group were referred to (for the combination of typical manifestations) as acrocephalosyndactyly.



Syndactyly in Apert syndrome

Genetics

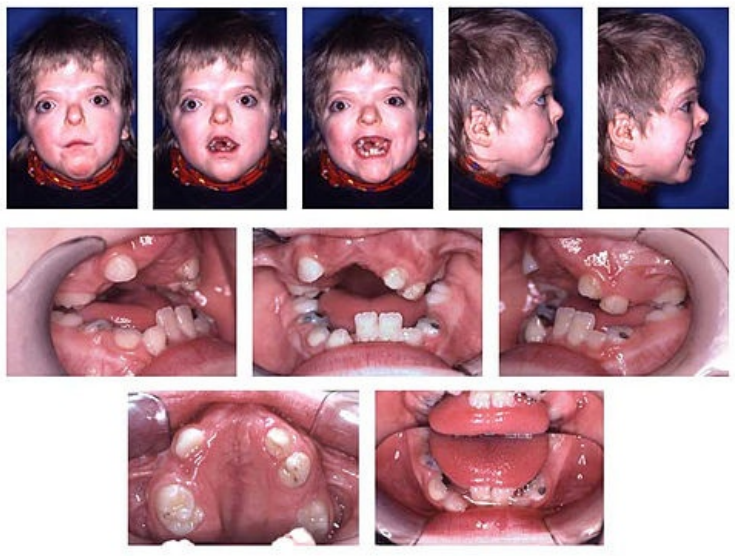
Apert syndrome is caused by a mutation in the gene for fibroblast growth factor receptor type 2 (*FGFR2*, 10q26.13, OMIM: 176943 (http://omim.org/entry/176943)). The inheritance of the syndrome is autosomal dominant.

Etiopathogenesis

- Probably a primary germ defect causing premature closure of sutures and fusion of fingers (syndactyly).

Clinical picture

- Elongation of the skull (turicephaly);
- syndactyly;
- multiple tarsal coalitions;
- deformities middle ear;
- cerebral atrophy (by the pressure of the growing brain on the rigid skeleton of the skull).



Appearance of a patient with Apert syndrome, dentitio tarda

Therapy

- Neurosurgical;
- orthopedic (disruption of cranial seams, separation of fingers).

Links

Related Articles

- Craniostenosis

References

- DUNGL, P., et al. *Ortopedie.* 1. edition. Prague : Grada Publishing, 2005. ISBN 80-247-0550-8.