Crouzon syndrome

Synonyms: Vogt's syndrome, dysostosis craniofacialis hereditaria, dysostosis cranio-orbito-facialis

Front view

The syndrome is named after the French physician Louis Edouard Octave Crouzon[1]

Genetics:

- OMIM: 123500 (https://omim.org/entry/123500)
- syndrome the syndrome is caused by a mutation in the gene for fibroblast growth factor receptor-2 (FGFR2; 10q26)
- Heredity: autosomal dominant
- There is also a rarer form of Crouzon syndrome with acanthosis nigrigancs (OMIM 612247 (https://omim.org/entry/612247)), which is due to a specific missense mutation in the FGFR3 gene in the region of 4p16.3.

Characteristics:

Side view

- early closure of cranial sutures
- expanded acrocephalic skull
- eye anomaly: exophthalmos, atrophy n. opticus, blindness, hypertelorism, divergent strabismus, nystagmus
- hypoplasia maxilla
- progressive intracranial hypertension

Links

References

1. OLE DANIEL, Enersen. *Whonamedit - Apert-Crouzon syndrome* [online]. [cit. 2011-04-24]. <http://www.whonamedit.com/synd.cfm/1383.html>.

References

- LAZOVSKIS, Ilmars – DOBIÁŠ, Václav. *Overview of clinical symptoms and syndromes.* 2. edition. Avicenum, zdravotnické nakladatelství, 1990. 0 pp. ISBN 80-201-0043-1.