

# Crouzon syndrome

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

The syndrome is named after the French physician Louis Edouard Octave Crouzon<sup>[1]</sup>

## 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 nigricans (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:

- 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

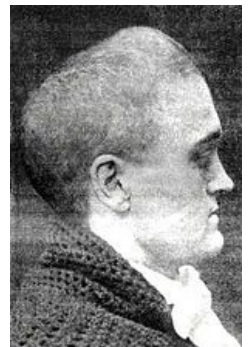
1. OLE DANIEL, Enersen. *Whonamedit - Apert-Crouzon syndrome* [online]. [cit. 2011-04-24]. <<http://www.whonamedit.com/synd.cfm/1383.html>>.

## References

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



Front view



Side view