Bloch-Sulzberg syndrome

Synonyms: Incontinentia pigmenti, melanoblastosis Bloch-Sulzberg, Siemens-Bloch-Sulzberg syndrome, melanoblastosis cutis linearis sive systematica, naevus pigmentosus systematicus.

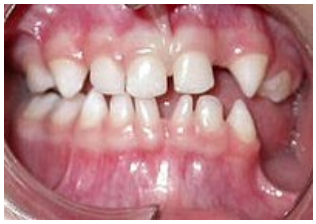
Named after Swiss dermatologist Bruno Bloch and American dermatologist Marion Baldur Sulzberg.[1]

Genetics:

- OMIM: 308300 (http://omim.org/entry/308300)
- The disease is caused by a mutation in the gene of *IKBKG* (*NEMO*) in the Xq28 region.
- Inheritance: X-linked dominant, male lethal, variable expression in females (due to random X-chromosome inactivation).

Characteristics:

- cinnamon dirty symmetrical skin spots on the sides,
- alopecia,
- dystrophy nails,
- defects of teeths,
- corneal opacity,
- microcephaly.



Deformation of teeth in a 3-year-old girl

Links

References

1. OLE DANIEL, Enersen. *Whonamedeit - Bloch-Sulzberger pigment dermatosis* [online]. [cit. 2011-04-25]. <http://www.whonamedit.com/synd.cfm/1762.html>.

References

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