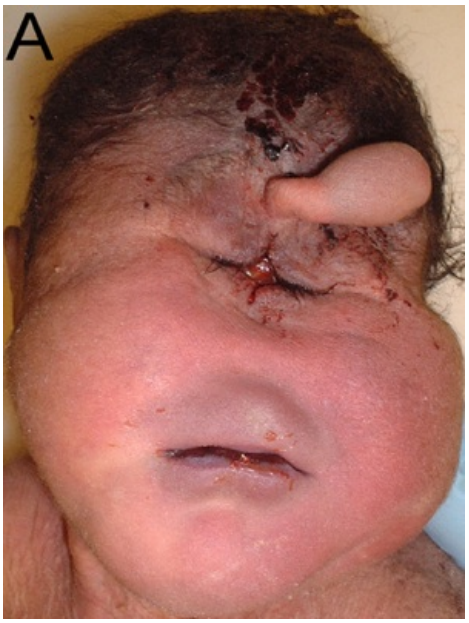
Holoprosencephaly

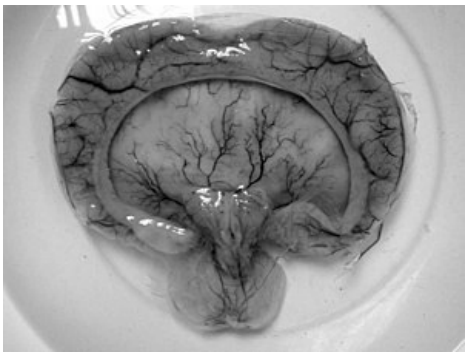
Holoprosencephaly (OMIM: 236100 (http://omim.org/entry/236100)) is a developmental defect characterized by the absence of central craniofacial structures, which has variable expressivity (only one front tooth, one common nasal cavity, one eye is present (**cyclopsism**); fusion of the cerebral hemispheres (**alobar holoprosencephaly**); or absent nervus olfactorius and tractus olfactorius and corpus callosum).



Cyclops (holoprosencephaly in Patau syndrome)

Causes of holoprosencephaly

1. Damage teratogens at the time of gastrulation, e.g. alcohol - selective killing of cells of central structures (at a time when the mother usually does not even know she is pregnant yet).
2. Patau syndrome or trisomy 18 (more rarely).
3. Poruchy biosyntézy cholesterolu (**Smith-Lemli-Opitz syndrom**), výsledkem je narušení signalizace SHH, pro kterou je cholesterol nezbytný.
4. Mutation of the gene SHH, which is expressed in the notochord, the prechordal plate, and also the base of the neural tube and affects the development of the forebrain (SHH induces the expression NKX2.1 for forebrain; SHH induces the expression of **PAX2'** inhibiting the effect of **PAX6** in the midline and thus initiating the division of the eye primordia and the adjacent region into paired).
5. Mutations of genes SIX3 (sine oculis homeobox), TGIF, ZIC2.


Alobar holoprosencephaly

Links

Used literature

- SADLER, Thomas W. *Langmanova lékařská embryologie : Překlad 10. vydání.* 1. edition. Grada Publishing, a.s, 2011. 432 pp. ISBN 978-80-247-2640-3.