Cri du chat syndrome

Template:Infobox - genetic disease **Cri du chat syndrome** *(cat cry/cat meow syndrome, monosomy 5p, Lejeune syndrome)* is one of the syndromes caused by a structural chromosomal aberration. The syndrome was named after the characteristic sound manifestations of affected newborns, whose **screams and cries resemble the meowing of a cat**.

Pathogenesis and frequency

Cri du chat syndrome is the first syndrome in which a structural chromosomal aberration[1] was described as a cause of the syndrom itself. This syndrome is specifically **a deletion** that occurs *de novo in 80% of cases*[2]. In 10–15% of cases[3], offspring of translocation carriers are affected. The locations of the breaks and the extent of the deleted area can be different for individual patients. **Deletion of the so-called critical region 5p15.2** (cri du chat critical region) is key to the manifestation of the syndrome. Due to the size of this region, the syndrome is classified among the microdeletion syndromes. The frequency of this disease in the population is 1/20,000[4]. Both males and females are affected.

Clinical picture and prognosis

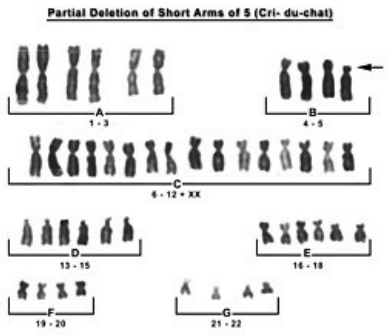
Newborns usually have a low birth weight and generally do not thrive. **Microcephaly** and various forms of facial deformities are evident (moon-shaped face, epicanths , hypertelorism, small lower jaw, malformations of the auricles). Congenital defects of internal organs, especially the heart, are also common. Individuals are significantly **psychomotor retarded**. The cry, reminiscent of a cat's meow, is caused by insufficient development of the larynx at an early age. Over time, this sound is modified. Affected people usually have a normal life expectancy. Early diagnosis allows the use of therapeutic methods for the development of mental and motor skills.

Diagnostics

Large deletions can also be detected by classical **cytogenetic examination**, but today the method of first choice (for any clinical suspicion of microdeletion syndrome) is **chromosomal microarray**. If there is a clear clinical suspicion of this particular syndrome, it is also possible to use **fluorescence in situ hybridization** (FISH) using a locus-specific probe.

In some cases, Cri du chat syndrome in the offspring may be the result of a balanced rearrangement affecting chromosome 5 in one of the parents, in which case the couple can be offered prenatal or even preimplantation genetic diagnosis.

The karyotype of the syndrome is 46,XX, del(5)(p?) or 46,XY, del(5)(p?) .
The question mark in the notation indicates the specific point of the break.



Cri du chat syndrome partial deletion of short arm of chromosome 5

Links

Related Articles

- Chromosomal abnormalities
- Numerical chromosomal abnormalities
- Syndromes due to aneuploidy of autosomes
  - Down syndrome
  - Edwards syndrome

External links

- Microdeletion syndromes, E. Seemanová (http://www.prolekare.cz/cas opis-lekaru-ceskych-clanek/mikrodelecni-syndromy-25743?confirm_rules=1)
- The cry of a child with Cri du chat syndrome (https://www.youtube.com/watch?v=TYQrzFABQHQ)

Used literature

- OTOVÁ, Berta, et al. *Lékařská biologie a genetika.* 2. edition. Praha : Karolinum, 2014. ISBN 9788024628356.
- SNUSTAD, D. Peter – SIMMONS, Michael J. *Genetika.* 1. edition. Brno : Masarykova univerzita, 2009. ISBN 9788021048522.
- NUSSBAUM, Robert L – MCINNES, Roderick R – WILLARD,, et al. *Klinická genetika: Thompson & Thompson.* 6. edition. Praha : Triton, 2004. 492 pp. ISBN 80-7254-475-6.

References

1. LEJEUNE, J – LAFOURCADE, J – BERGER, R. , et al. [3 CASES OF PARTIAL DELETION OF THE SHORT ARM OF A 5 CHROMOSOME]. *C R Hebd Seances Acad Sci* [online]*.* 1963, vol. 257, p. 3098-102, Available from <https://www.ncbi.nlm.nih.gov/pubmed/14095841>. ISSN 0001-4036.
2. NIEBUHR, E. *Medscape* [online]. [cit. 2016-09-24]. <https://emedicine.medscape.com/article/942897-clinical>.
3. NUSSBAUM, Robert L – MCINNES, Roderick R – WILLARD, Huntington F, et al. *Klinická genetika: Thompson & Thompson.* 6. edition. Praha : Triton, 2004. 492 pp. pp. 166–167. ISBN 80-7254-475-6.
4. **Cite error: Invalid <ref> tag; no text was provided for refs named 0tová**