

Beckwith - Wiedemann Syndrome

Beckwith-Wiedemann syndrome (BWS) also **EMG syndrome** (exomphalos, **macro**glossia, **gig**antism) is a syndrome associated with excessive growth and an increased risk of tumors. It is associated with dysregulation of imprinting of one of the two groups of imprinted genes. The best studied is the region including two reciprocally imprinted genes – the gene for the growth factor IGF2, expressed from the paternal allele, and the gene **H19**, expressed from the maternal allele. In patients with BWS, paternal duplications are found on the short arms of chromosome 11 (region **11p15**), paternal UPD, deletion or translocation of the maternal allele H19 (the consequence is the activation of the maternal allele IGF2), or mutation or deletion of the controlling element, the so-called imprinting center. All this leads to excessive production of the IGF2 product and therefore to manifestations of excessive growth and the risk of tumors. A proportion of BWS patients have dysregulation of the second imprinted region at 11p15, where several genes are located, including the maternally expressed gene for the cyclin dependent kinase (CDK) inhibitor.

Epidemiology

- Incidence worldwide¹ : 13 700.
- more often during in vitro fertilisation.
- Incidence is not tied to race or gender.^{[1][2][3]}

Clinical Picture

- Macrosomia
 - rapid growth at the end of fetal development and in the first years of life, normal growth in adulthood
 - macroglossia, hemihypertrophy
 - visceromegaly – hepatomegaly, nephromegaly (the spleen, pancreas, adrenal glands may also be enlarged).
- Defects of the anterior abdominal wall (omphalocele, umbilical hernia, diastasis of rectus abdominis muscles)
- Neonatal hypoglycemia (30–50 % of children with BWS).
- The typical shape of the tragus - 'notches'
- Polyhydramnios, large placenta
- Fetal adrenocortical cytomegaly
- Kidney anomalies – malformations, renal medullary dysplasia, nephrocalcinosis, nephrolithiasis.
- Wilms tumor (in 5–7 % of children with BWS), hepatoblastoma, less commonly neuroblastoma, rhabdomyosarcoma, adrenocortical carcinoma.^{[1][2][3]}

Diagnostics

- Clinical diagnosis according to typical phenotypic manifestations.
- Verification of the clinical diagnosis by targeted molecular genetic examination of the critical region at 11p15 (the examination is also available in the Czech Republic)

Care of patient with BWS

- Maintaining euglycemia.
- frequent screening USG of the abdomen (every 4 months), alpha-fetoprotein level (hepatoblastoma screening), dispensary by a pediatric oncologist
- tongue resection for DC obstruction (rarely necessary)
- Monitoring growth and development^{[1][2][3]}

Links

Related Articles

- Silver-Russell syndrome
- Gene imprinting
- Prader-Willi syndrome
- Angelman syndrome
- Uniparental disomy

External links

- Beckwith-Wiedemann Syndrome – eMedicine (<https://emedicine.medscape.com/article/919477-overview>)
- OMIM:130650 (Beckwith-Wiedemann Syndrome) (<http://omim.org/entry/130650>)

Source

- POLÍVKOVÁ, Z. Imprinting genů a lidské patologie. *Čas. Lék. čes.* 2005, y. 144, vol. 4, p. 245-250, ISSN 1803-6597.

Reference

1. FERRY, Robert J., Jr. *Beckwith-Wiedemann Syndrome* [online]. [cit. 2012-02-04]. <<https://emedicine.medscape.com/article/919477-overview#showall>>.
2. Online Mendelian Inheritance in Man®. *Beckwith-Wiedemann Syndrome; BWS* [online]. [cit. 2012-02-04]. <<https://omim.org/entry/130650#contributors-shutter>>.
3. WIKIBOOKS. *Handbook of Genetic Counseling : Beckwith-Wiedemann Syndrome* [online]. [cit. 2012-02-04]. <https://en.wikibooks.org/w/index.php?title=Handbook_of_Genetic_Counseling/Beckwith-Wiedemann_Syndrome&oldid=491429>.