

Beckwith-Wiedemann Syndrome

English: *Beckwith-Wiedemann syndrome*



- This uniparental disomy is connected with **changes of methylation on chromosome 11 (11p15)**. The syndrome affects more parts of the body.
- *Typical symptoms:* macrosomia (overgrowth or simply gigantism), omphalocele (or related abdominal wall defect), large tongue (macroglossia), kidney abnormalities, hypoglycemia (because of the hyperinsulinisms) and increased risk of tumors (like Wilms tumor) during the childhood. About 20% of patients die in early age because of the complications of these congenital symptoms.
- It is also called **EMG syndrome** (exomphalos, **m**acroglossia, **g**igantism).
- Incidence of Beckwith-Wiedemann syndrome is *1 in 12 000 people* worldwide.
- Molecular-genetic diagnostics (including the identification of abnormal methylation) is possible.

Links

Related articles

- Genomic Imprinting
- Gene
- Allele
- Chromosome

Sources

- What are Genomic Imprinting and Uniparental Disomy (<http://ghr.nlm.nih.gov/handbook/inheritance/updimprinting>)
- What is Genomic Imprinting? (<http://www.geneimprint.com/site/what-is-imprinting>)

Bibliography

- KUMAR, ABBAS, FAUSTO, MITCHELL,. *Robbins Basic Pathology*. 8th edition edition. 2007. ISBN 978-0-8089-2366-4.