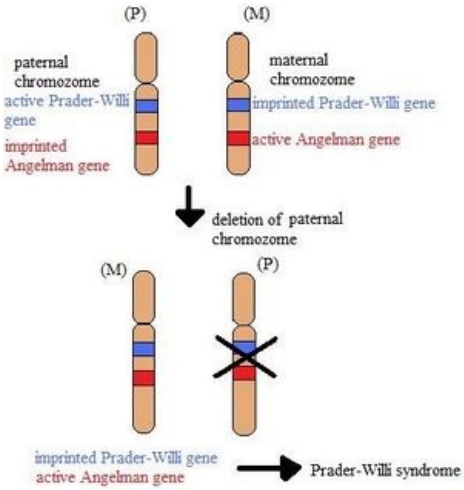
Prader-Willi Syndrome

English: *Prader-Willi syndrome*

- About 2/3 of cases are induced by **the loss of paternal chromosome 15** segment (The Prader-Willi/Angelman Critical Region- PWACR; 15q11-13). Physiologically, just genes in the paternal region are functional (the maternal copies are imprinted, i.e. epigenetically silenced) – opposite to Angelman syndrome.
- *Typical symptoms*: hypotonia, obesity, poor growth, delayed development and higher appetite (*hyperphagia* – leads to overeating).
- Children have also problems with their learning abilities and behavior. We can also find typical physical features – smaller feet and hands, fair skin, underdeveloped genitals (*hypogonadism*). Some of the affected patients are infertile.
- Incidence of Prader-Willi syndrome is *1 in 10,000–30,000* people worldwide.



Chromosome 15 – **Prader-Willi syndrome**

Links

Related articles

- Genomic Imprinting
- Angelman syndrome
- 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.