

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.

Links

Related articles

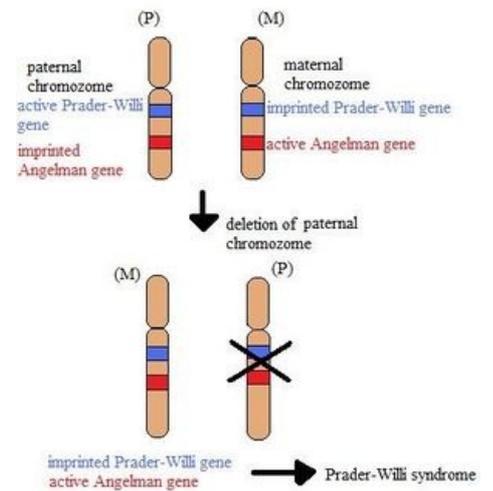
- Genomic Imprinting
- Angelman syndrome
- Gene
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- Chromosome

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.



Chromosome 15 - Prader-Willi syndrome