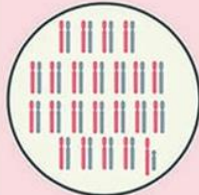


# Синдром Прадера-Вилли

Лысенко Анна, 3 курс, 11001616

# GENETIC CAUSES OF PRADER-WILLI SYNDROME



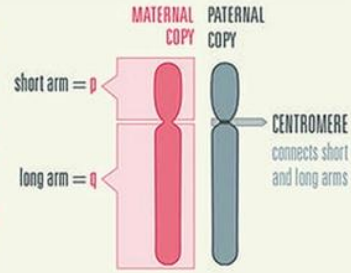
EVERY CELL HAS **23 PAIRS** OF CHROMOSOMES

**22 PAIRS** OF NUMBERED CHROMOSOMES AND **ONE PAIR** OF **SEX** CHROMOSOMES

EACH PAIR HAS A **MATERNAL** AND **PATERNAL** COPY

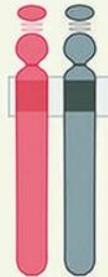


EACH COPY HAS 2 ARMS



**PRADER - WILLI**  
**CRITICAL REGION**  
**LOCATED ON**  
**CHROMOSOME 15**

15



LABELING CONVENTIONS  
"GENE ADDRESS"  
ARM  
15q11 - q13  
CHROMOSOME LOCATION ON ARM

**NORMAL**  
CHROMOSOME 15

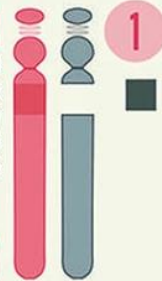


**LOSS** OF GENE FUNCTION IN  
**15q11-q13** REGION ON  
**PATERNAL** CHROMOSOME  
**CAUSES PRADER-WILLI**

## 3 GENETIC MANIFESTATIONS OF PWS

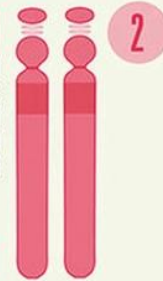
**DELETION**  
non-inherited  
deletion in the  
paternally  
contributed  
chromosome 15

**70%**



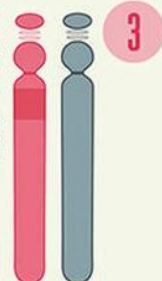
**UPD**  
maternal  
uniparental disomy:  
two maternal  
and no paternal  
chromosome 15

**28%**



**GENETIC**  
**IMPRINTING**  
paternal contribution  
is non-functional

**2%**









Body habitus, facial and hand dysmorphism in molecularly confirmed DWS cases



Narrow temple distance  
and nasal bridge

Almond-shaped eyes  
Mild strabismus

Thin upper lip  
Downturned mouth

Overweight

# Prader Willi Syndrome



Genetic disorder due to loss of genes on chromosome 15



1<sup>st</sup> described in 1887 by John Langdon Down



Named after Andrea Prader, Heinrich Willi & Alexis Labhartin 1956



70% cases are inherited from father



Affects 1 in 20,000 people



Symptoms are weak muscles, poor feeding, physical defects & delayed development



Diagnosed by physical examination & genetic analysis



No curative treatment available



Drugs, hormonal therapies & supportive care improve outcome



Complications are type 2 diabetes, obesity, intellectual defects & infertility

