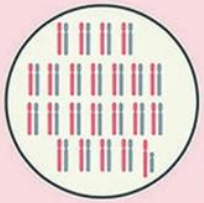
Синдром Прадера-Вилли

Лысенко Анна, 3 курс, 11001616

GENETIC CAUSES OF PRADER-WILLI SYNDROME

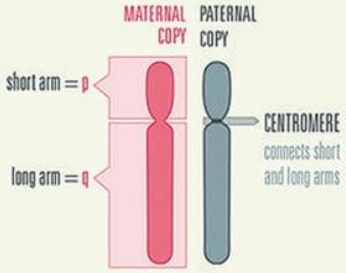


EVERY CELL HAS 23 PAIRS OF CHROMOSOMES

22 PAIRS OF NUMBERED CHROMOSOMES AND ONE PAIR OF SEX CHROMOSOMES

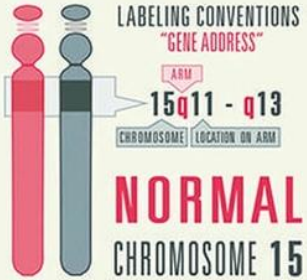
EACH PAIR HAS A MATERNAL AND PATERNAL COPY

EACH COPY HAS 2 ARMS



PRADER - WILLI CRITICAL REGION LOCATED ON CHROMOSOME 15

15



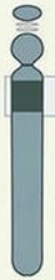
LABELING CONVENTIONS "GENE ADDRESS"

ARM

15q11 - q13

CHROMOSOME | LOCATION ON ARM

NORMAL CHROMOSOME 15

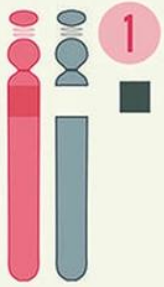


LOSS OF GENE FUNCTION IN 15q11-q13 REGION ON PATERNAL CHROMOSOME CAUSES PRADER-WILLI

3 GENETIC MANIFESTATIONS OF PWS

DELETION 1

non-inherited deletion in the paternally contributed chromosome 15



70%

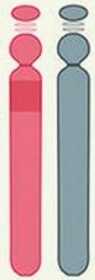
UPD 2

maternal uniparental disomy : two maternal and no paternal chromosome 15

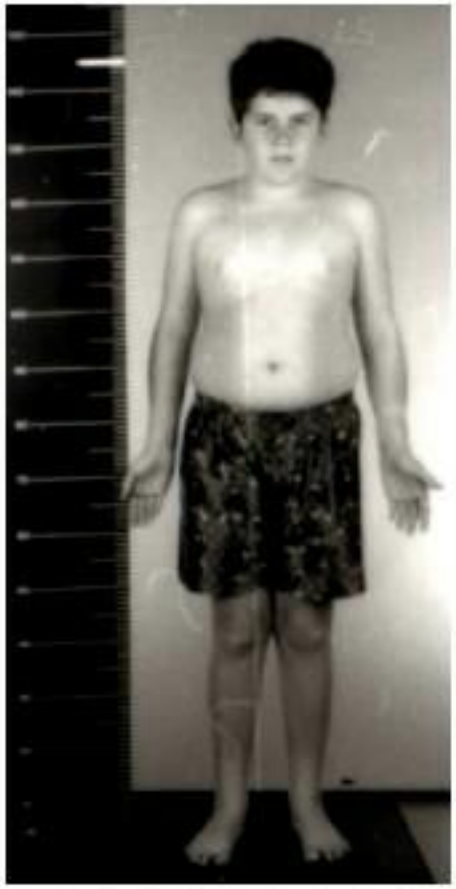
28%

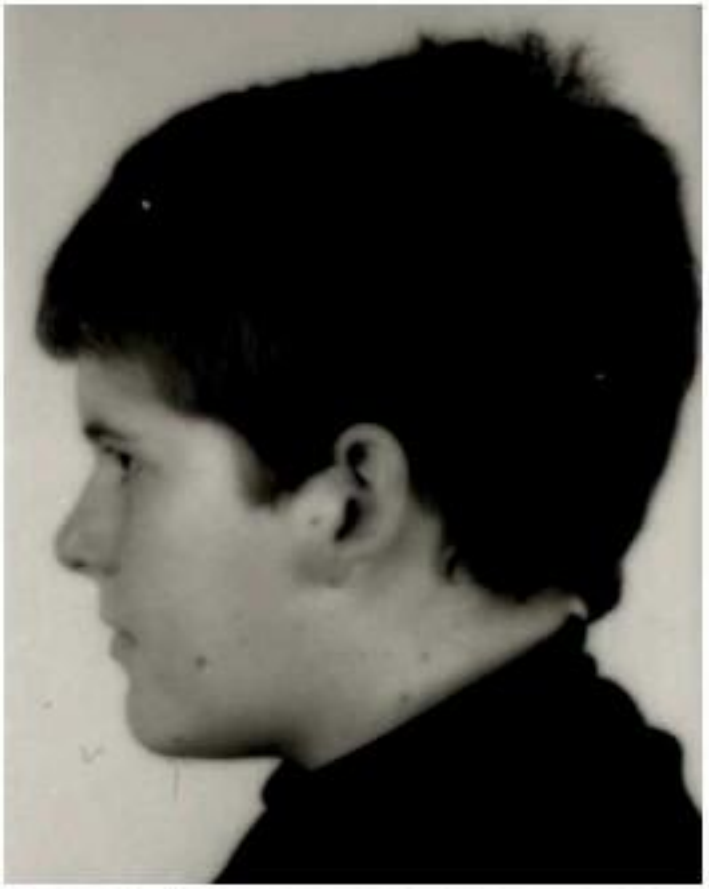
GENETIC IMPRINTING 3

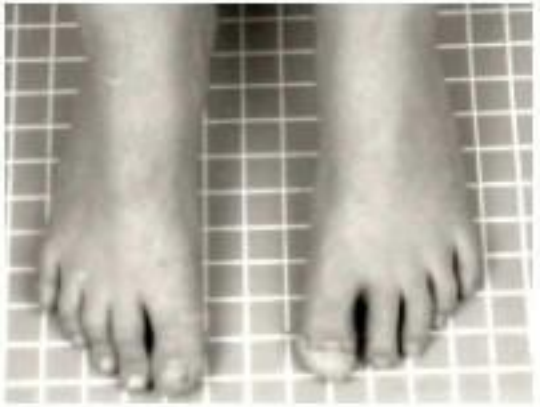
paternal contribution is non-functional

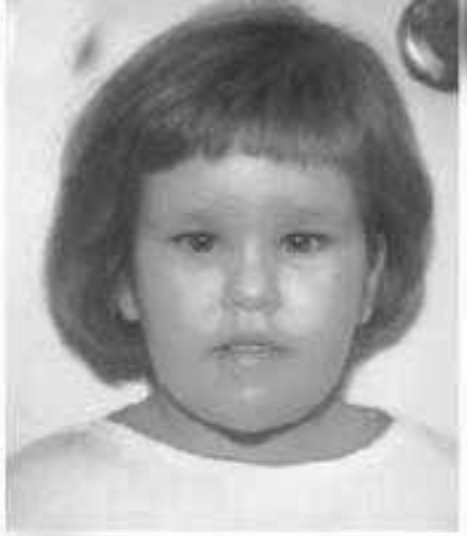
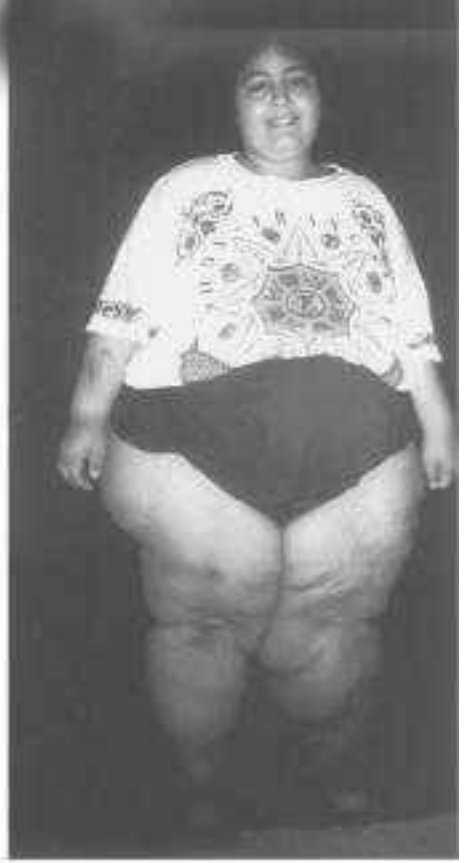


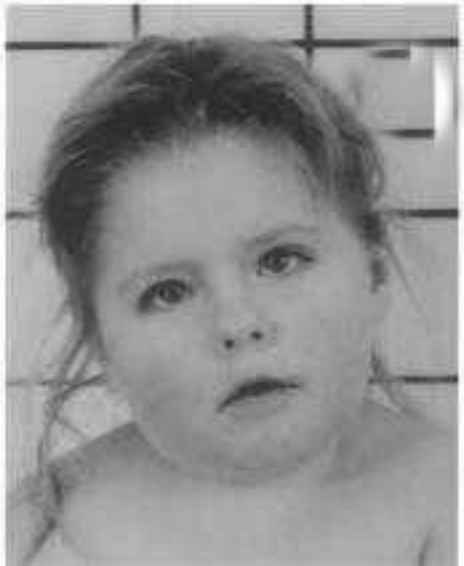
2%

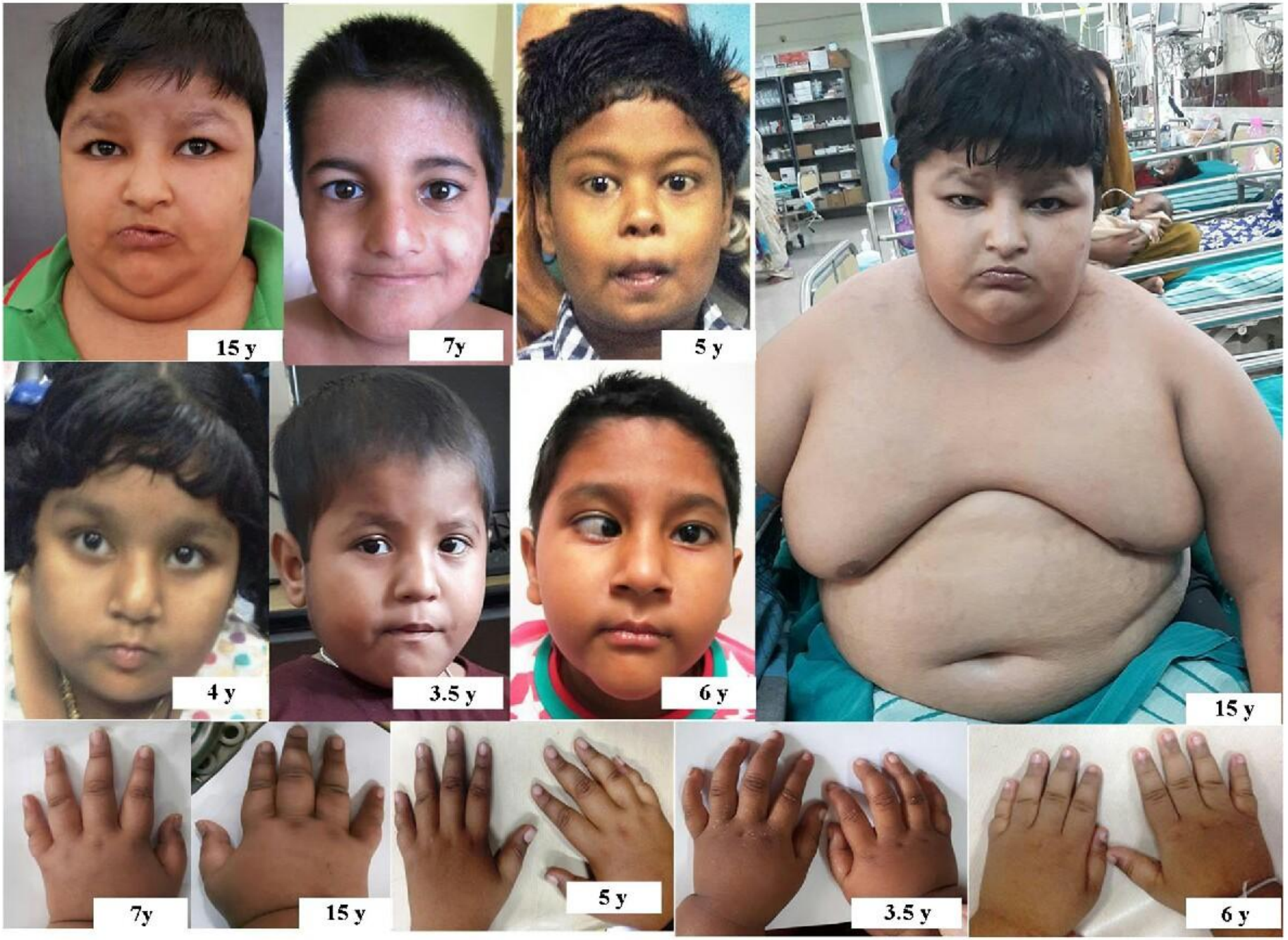




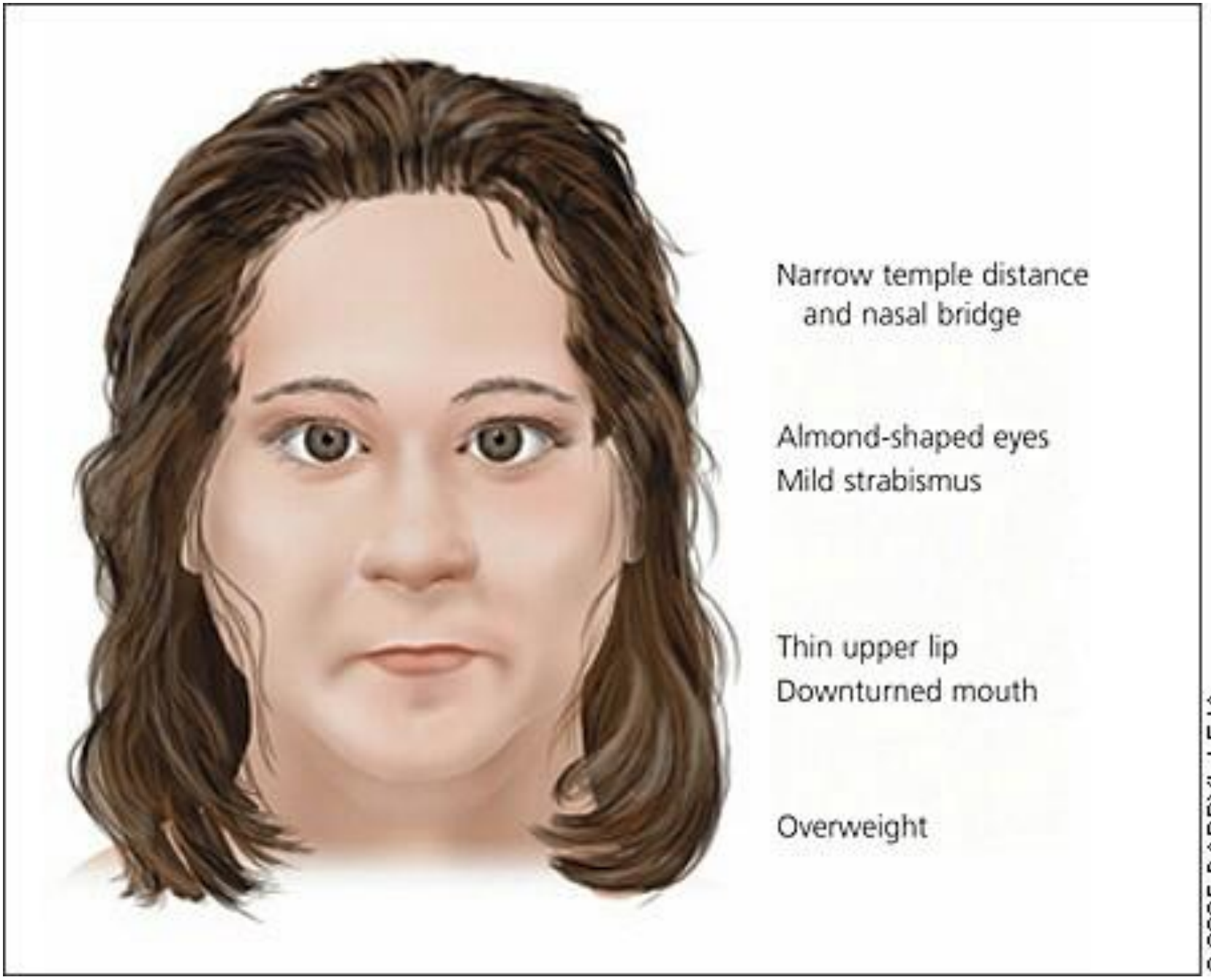





Body habitus, facial and hand dysmorphology in molecularly confirmed PWS cases



Prader Willi Syndrome

XpertDox
Clinical, Education, Research

Genetic disorder due to loss of genes on chromosome 15

1st 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