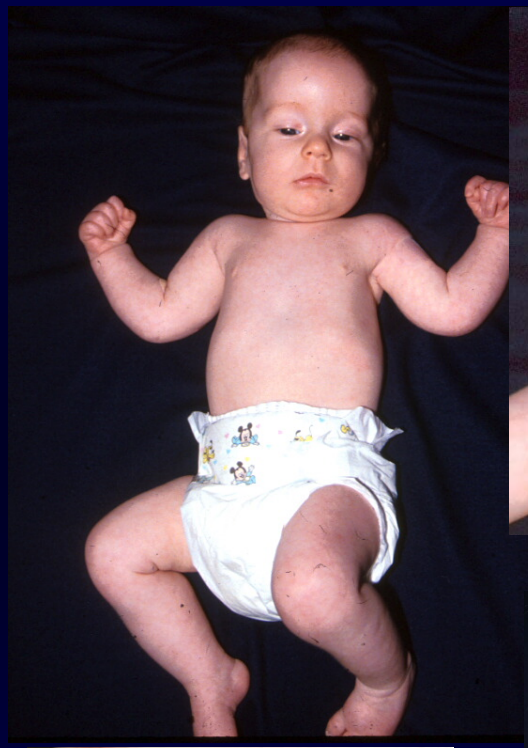
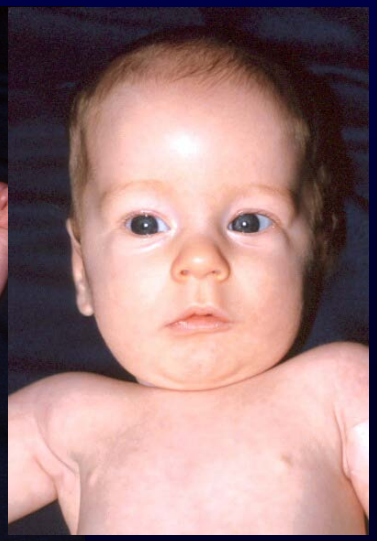
Prader-Willi Syndrome

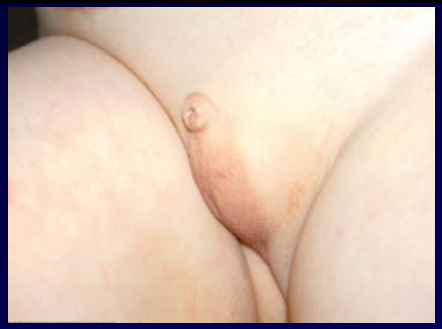
- Neonatal muscular hypotonia
- FTT, poor feeding in infancy
- Obesity in childhood, hyperphagia
- Hypogenitalism
- Short stature, small hands and feet
- Intellectual deficiency and behavioral problems

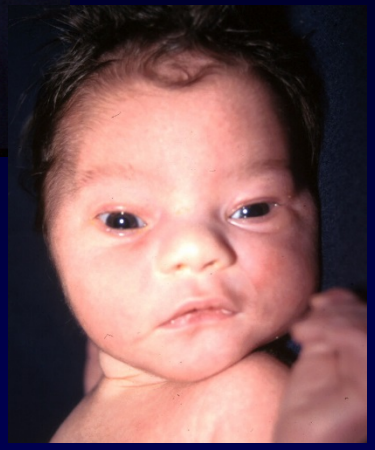
- Etiology: most due to paternal-derived 15q11-13 deletions (detected by chromosome microarray or FISH and DNA methylation testing) or maternal uniparental disomy (UPD) of chromosome 15.

Neonatal



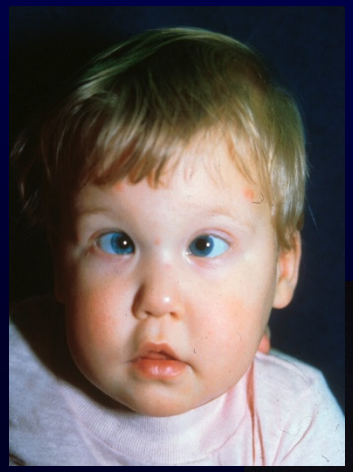


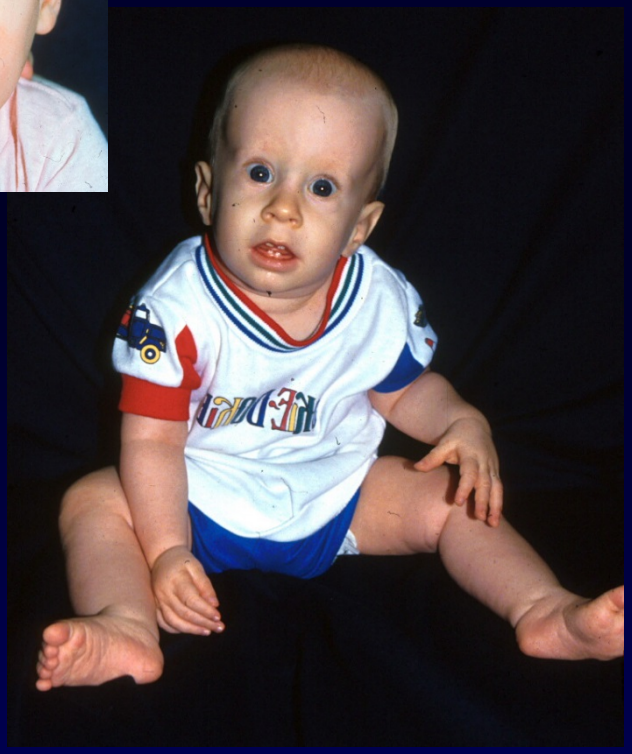




- Low muscle tone
- Poor suck, FTT
- Hypogonadism
- Relatively larger forehead
- Narrow bifrontal area
- Down-turned mouth
- Almond-shaped eyes

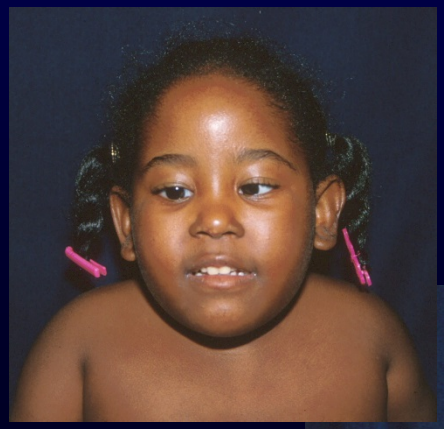
Infancy

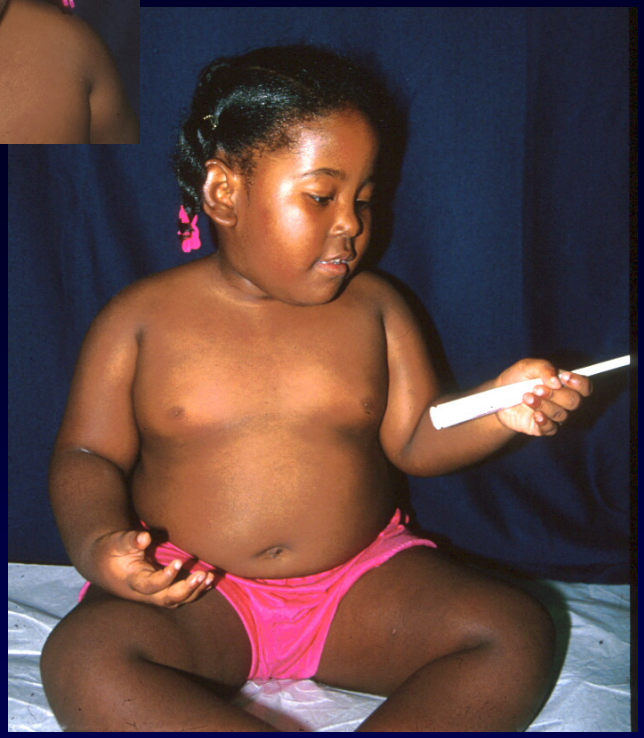




- Developmental delay is noted
- Hypotonia improves
- Strabismus may occur
- Feeding and activity improves

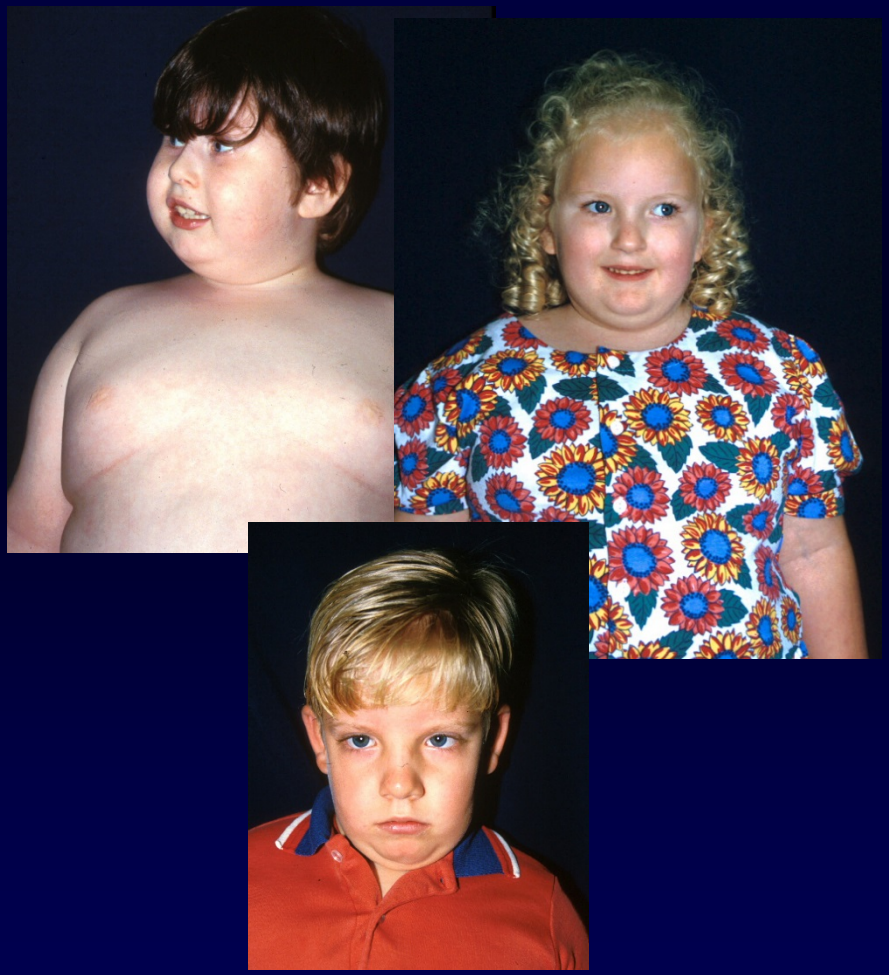
Age 1-3 years





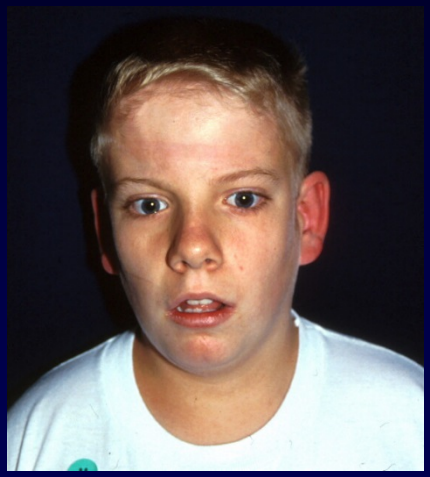
- Obesity
- Hyperphagia
- Small hands and feet
- Developmental delay
- Behavioral disturbances

Early Childhood



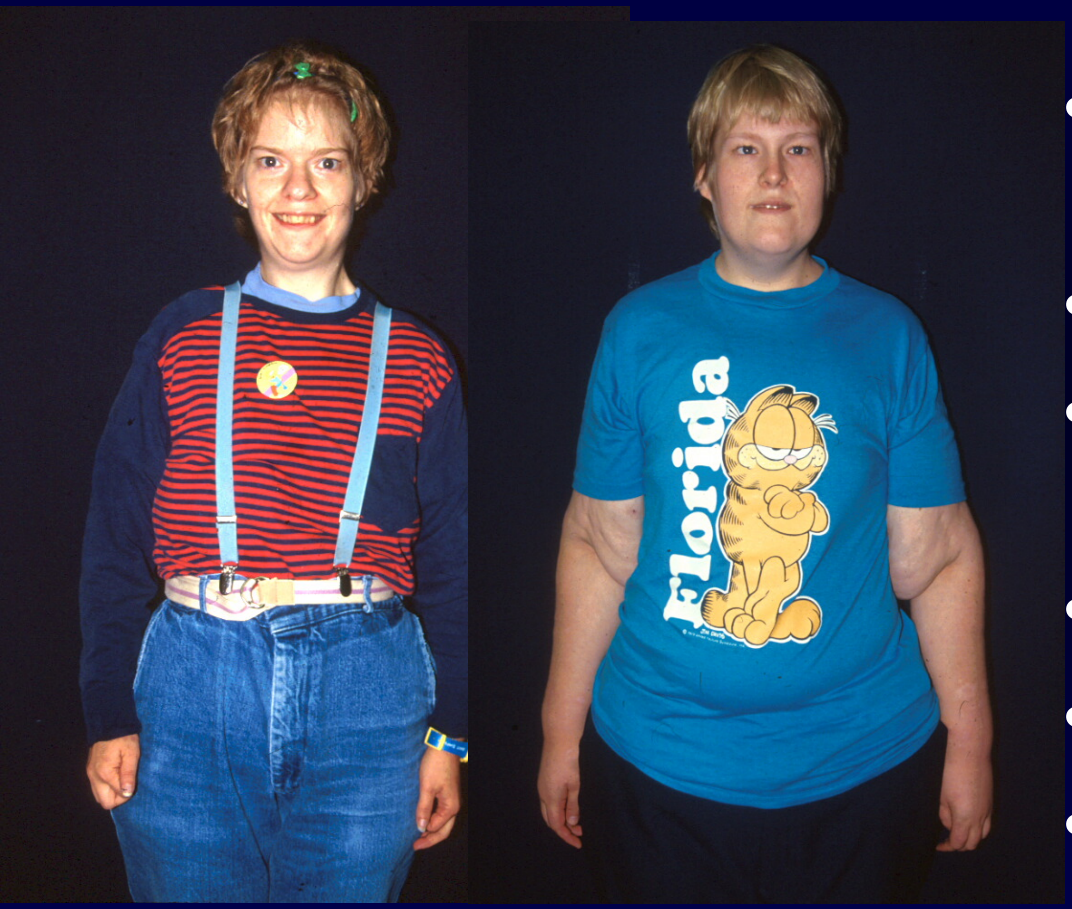
- Obesity
- Voracious appetite
- Skin picking
- Small hands and feet
- Behavioral changes
- Developmental delay
- Short stature
- Small genitalia

Older Children, Teenagers



- Obesity
- Behavioral Issues
  - Temper tantrums
  - Obsessive -compulsive behavior
  - Stubbornness
  - Self-injurious behavior
- Sexuality
- Relatively mild intellectual deficiency

Adults

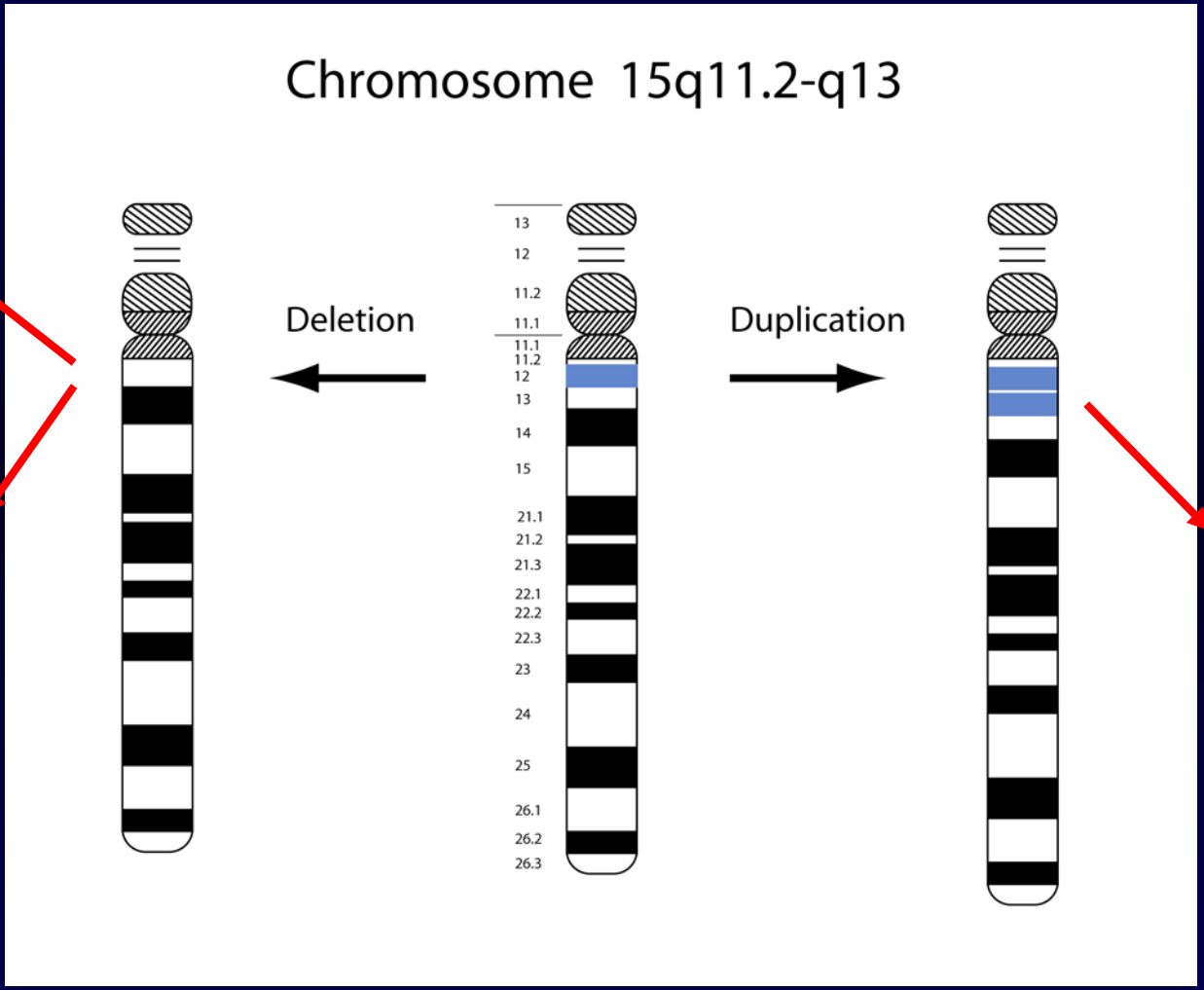


- Semi-independent living
- Obesity control
- Behavioral management
- Social involvement
- Vocational training
- Medical issues

Deletion on the paternally-derived 15 can cause Prader-Willi syndrome

Angelman
Maternal 15

Prader-Willi
Paternal 15



Autism-like
Maternal 15