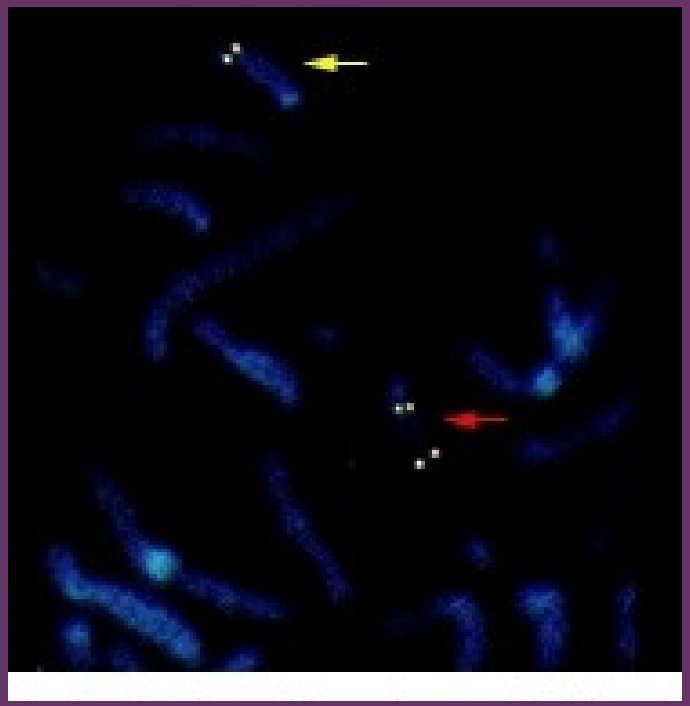


# Prader Willi Syndrome

Annie Smartt

# What is Prader Willi Syndrome

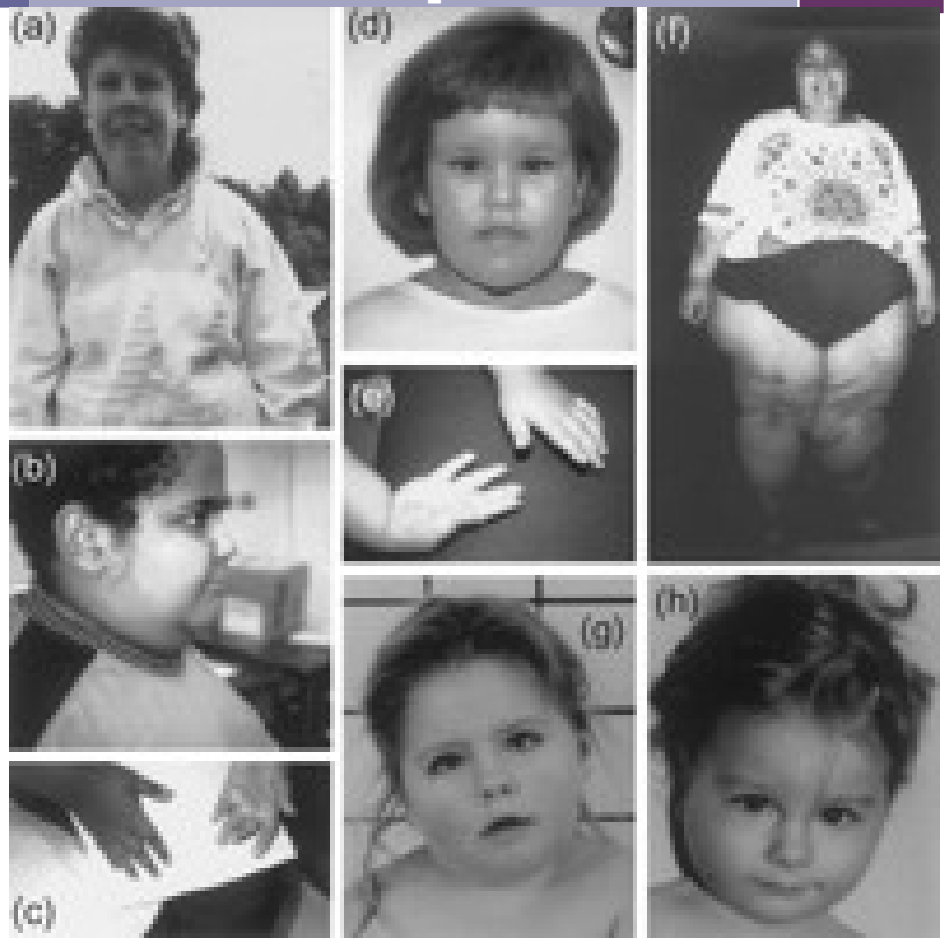
- Uncommon genetic condition
- Caused by absence of segment 11-13 on chromosome 15
- Characterized by mental retardation, hypotonia, short stature, hyperphagia, hypogonadism, obesity
- 1/10,000 – 1/30,000

# Classical Diagnosis

## Consensus Diagnostic Criteria

- Neonatal/infantile hypotonia with poor suck
- Feeding problems
- Onset of rapid weight gain causing central obesity
- Hyperphagia
- Characteristic facial features
- Hypogonadism – delayed puberty, infertility
- Developmental delay/ learning disabilities

## Examples



# Manifestations

- Obesity = greatest problem to health
- Diabetes
- Skin problems e.g. chronic edema
- Behavioral and Psychiatric problems:
  - Temper tantrums
  - Stubbornness
  - Manipulative Behaviors

# Treatment Options

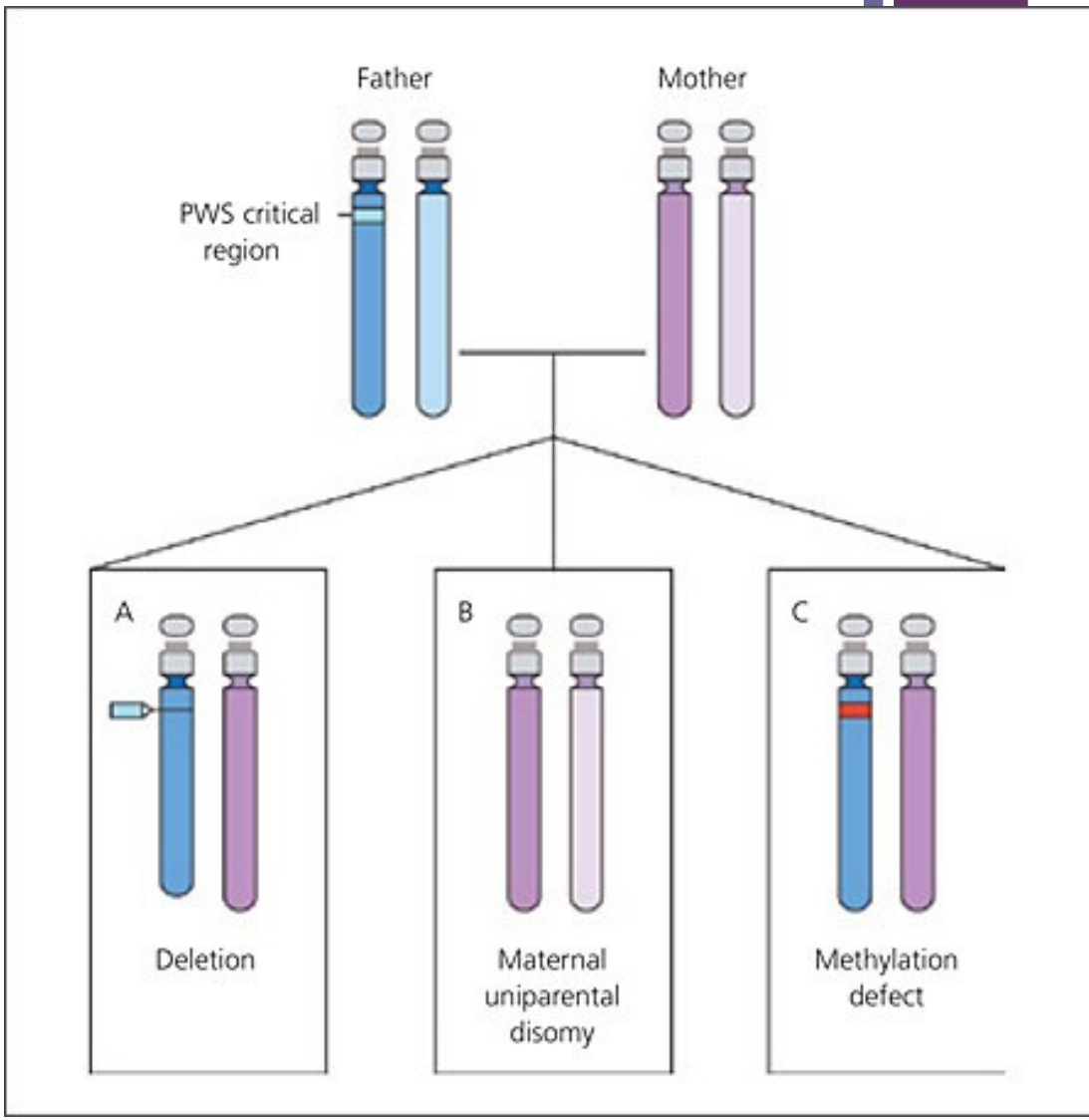
- Feeding techniques
- Physical Therapy
- Low calorie diet/exercise plan
- Human Growth Hormone
- Serotonin reuptake inhibitors

# Genetic Explanation

- Epigenetics: modification of activation of certain genes but not basic structure of DNA
- Genomic Imprinting: preferential expression of a gene from only one of the two parental alleles
- Not Mendelian Genetics
- PWS caused from loss of paternally inherited genes on chromosome 15q11.2-q13

# Genetics Continued

- Causes of Prader Willi
  - 70% Deletion of paternal chromosome 15
  - 29% Maternal Uniparental disomy
  - 1% Imprinting defect



# Even More Genetics

## Explanation

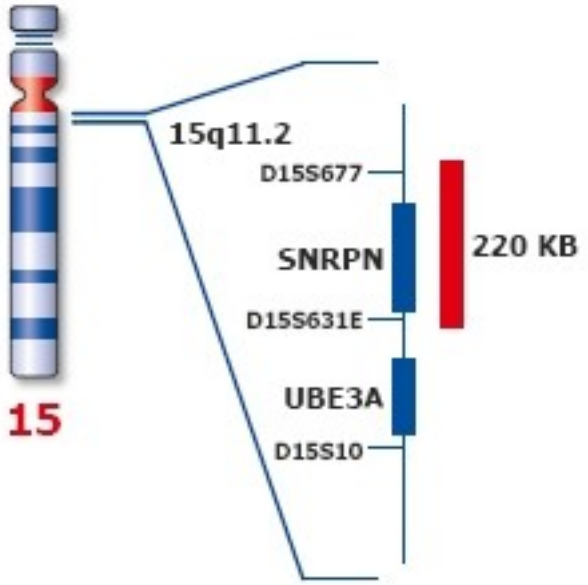
- Random genetic changes – not usually inherited
- No predisposition
- Parents unaffected
- Risk to sibling based on underlying cause of lack of expression
- Complete penetrance

## Better Diagnostics

- Molecular Genetic Testing
- Methylation analysis
- FISH
- Uniparental Disomy studies

# Better Therapies?

- Not at this time
- Irreversible condition
- Studies  - manifestations not cause
- Prenatal diagnoses



# Resources

- Gene Reviews, Prader Willi Syndrome
- Medline Plus, Prader Willi
- OMIM, Prader Willi Syndrome
- Diseases Associated with Imprinted Genes
- NCBI
- Genetics Home Reference