

15) Prader Willi Syndrome (PWS)



❖ Clinical features in infancy

- ✓ Hypotonia
- ✓ feeding problems

❖ Clinical features in childhood

- ✓ Hyperphagia
- ✓ Central obesity
- ✓ Hypogonadism
- ✓ Short stature
- ✓ Small hand and feet
- ✓ Mild intellectual disability
- ✓ Almond-shaped eyes
- ✓ Behavioral disorder



It is the most common syndromic form of obesity

❖ Which medication had FDA approval to be used with PWS?

- ✓ Growth hormone therapy

❖ Why they develop short stature, central obesity & hypogonadism ?

- ✓ Secondary to Hypothalamic and pituitary dysfunction

❖ Mode of inheritance?

- ✓ Sporadic
- ✓ Paternal derived deletion 15q11–13 (80%)
- ✓ Maternal UPD (20%)

Management Of Prader Willi Syndrome

JA Banks



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