

# Klinefelter Syndrome 47, XXY

- Incidence 1:800 males
- Neonates and infants can be asymptomatic.
- Later childhood:
  - Delayed sexual development
  - Hypogonadism, hypogonadism
  - Relatively long limbs
  - Intellectual deficiency, behavioral problems
  - Gynecomastia
- Often detected coincidentally as is also seen in the 47, XYY and 47, XXX condition.



Normal infant, diagnosed coincidentally by amniocentesis for advanced maternal age.

47, XXY in boy referred for evaluation of learning delay (left),  
and in a teenager with relatively long limbs, delayed  
secondary sexual traits and small testes.



- More than 2 extra X chromosomes cause progressive severity in intellectual deficiency and dysmorphic traits.

XXXY

XXXY

XXXXY

- However, these conditions occur much more infrequently than is observed for the XXY condition.