WISCONSIN BIRTH DEFECT PREVENTION AND SURVEILLANCE SYSTEM
DOWN SYNDROME

- Incidence 1/800
- Hypotonia

Dysmorphology
- Flat Facial profile
- Upslanting palpebral fissures
- Congenital Heart Disease
- Intestinal problems
- Mild to moderate CI
DOWN SYNDROME KARYOTYPE
Trisomy 13

Cleft lip and palate

- Cleft lip and palate
- Punched out scalp lesion
- Microphthalmia
- Polydactyly
Karyotype from a female with Patau syndrome (47,XX,+13)
Trisomy 18

- Microphthalmia
- Low set ears
- Congenital heart defects
- Rocker bottom feet
Trisomy 18 Karyotype
NOONAN SYNDROME

- Excess nuchal skin
- Developmental delay
- Pulmonic stenosis
- Short stature
Turner Karyotype
TURNER SYNDROME

Cystic hygroma

Excess nuchal skin
Clinical Features of Turner Syndrome

• Short stature
• Gonadal dysgenesis
• Ovarian Failure
• Coarctation of the aorta
• Renal anomalies
• Lymphedema
KLINEFELTER SYNDROME

Tall stature

Hypogonadism

Developmental delay
Klinefelter Syndrome

- 47,XXY
- Incidence 0.2%
- Other forms:
  - 48,XXXXY
  - 49,XXXXXY
  - 46, XY + Y sequences

Photo courtesy of KS & A
Clinical Features

- Testicular dysfunction
- Elevated urinary gonadotropins
- Microorchidism
- Eunochoidism
- Azoospermia
- Gynecomastia

Photo courtesy of KS&A
Potter Syndrome

• Renal Agenesis
• Pulmonary hypoplasia
• Death due to respiratory complications
Achondroplasia
22q Deletion

- Cleft palate
- Hypocalcemia
- Conotruncal heart defects
- Immune deficiency
PRADER-WILLI SYNDROME

Hypotonia

Failure to thrive

Hyperphagia