Down Syndrome

- Most common chromosomal condition
- **~1/800 live births**
- Leading genetic cause of intellectual disability
- Every child with Down syndrome is **unique**!

Etiology

- **Trisomy 21 (94%)**
  - Meiotic nondisjunction
- **Robertsonian translocation (3.3%)**
  - Potentially heritable
- **Mosaicism (2.4%)**
  - Milder phenotype

Risk Factors

- Maternal age > 35 years
- Previous pregnancy with chromosomal condition
- Pre-natal screening
  - Nuchal translucency
  - Absent nasal bone
  - Biochemical markers

Diagnosis can be either **pre-natal** (chromosomal karyotype via CVS or amniocentesis) or **post-natal** (chromosomal karyotype if ≥ 2 dysmorphic features seen)

Presentation

Classic Features

- **Craniofacial**
  - Brachycephaly, upslanted palpebral fissures, epicanthal folds, Brushfield spots, midface hypoplasia, flat/short nose, macroglossia, dental hypoplasia, protruding helix, short neck
- **Hands**
  - Single transverse palmar crease, broad fingers, clino/brachydactyly, small 5th mid-phalanx
- **Feet**
  - ↑ space between 1st + 2nd toes
- **General**
  - Hypotonia, hyperflexibility

Complications

- **OPT:** cataracts (✓ red reflex), strabismus
- **HEENT:** hearing loss, recurrent AOM
- **DENT:** periodontal disease
- **CV:** cardiac anomaly (50%)
  - AVSD > VSD, ASD, TOF
- **RESP:** OSA, pulmonary HTN
- **GI:** duodenal atresia, Hirschsprung, Celiac
- **GU:** cryptorchidism, male sterility
- **MSK:** C1/2 instability, hip dysplasia
- **CNS:** development delay, ASD, early Alzheimer's
- **ENDO:** hypothyroid, obesity, short stature
- **HEME:** leukemia (AML, ALL, transient)

Management

Surveillance

- **GENERAL:** T21 specific growth charts, routine immunizations + RSV (1st yr of life)
- **OPT:** newborn test by 3mo, then yearly
- **HEENT:** newborn hearing screen + behavioral test q6mo until 3yo, then yearly
- **DENT:** consult by 2yo, then every 6mo
- **CV:** newborn echo + peds cardio consult
- **RESP:** OSA monitoring, PSG by 4yo
- **GI:** constipation Tx, Celiac screen at 2yo
- **MSK:** observe for spinal cord compression
- **ENDO:** newborn TSH/T4, then yearly
- **HEME:** newborn CBCd, then CBC yearly

Holistic Care

- Early lactation support
- Referral to allied health services by 3yo
  - PT/OT/SLP
- Consult teachers to support individualized learning needs
- Mental health support
- Parental support
  - Community resources
  - Advocacy groups
  - Financial assistance

Use Appropriate Language

1. Down *not* "Down's"
2. Use person-first language
3. Be positive
4. End the R-word

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