1 Craniofacial Syndromes

2 Treacher Collins
   • Malar hypoplasia (81%)
   • downsllanting palpebral fissures (89%)
   • defect of lower lid (coloboma) (69%)
   • malformation of external ear 77%

3 Other sx’s
   • mandibular hypoplasia (78%)
   • partial absense eyelashes (53%)
   • ear canal defect 36%
   • conductive hearing loss 40%
   • cleft palate 28%
   • Scalp hair on cheek (26%
   • MR - rare 5%
   • cosmetic improvement with plastic surgery
   • inherited
   • wide variance in expression

4 Apert’s Syndrome (Acrocephalosyndactyly)
   • www.worldcf.org/cran (World Craniofacial Foundation
   • relatively uncommon - 1/160,000
   • +/- family history
   • recurrence risk for unaffected parents - minimal kid’s of Apert’s - 50% recurrence risk

5 Phenotype
   • tower shaped skull 2ndary to craniostenosis
   • under-developed mid face; recessed cheekbones and prominent eyes
   • syndactyly
   • +/- hypertelorism
   • +/- cleft
   • +/- MR

6 Apert’s

7 Crouzon’s Syndrome
   • craniofacial dystosis
   • rare; related to Apert’s
   • 1/10,000
   • Three distinct features:
     – abnormal shaped skull 2ndary to craniostenosis
     – underdeveloped midface & exophthalmos
     – ocular proptosis (eye prominence 2ndary to shallow orbits)
Mom and Son

Other sx’s
- hypertelorism
- parrot-like nose
- inverted V shape to palate
- conductive hearing loss

Robin Sequence
- multiple anomaly syndrome
- results from sequence-one deformity relates to occurrence of another
- occurs at 9 weeks gestation
- phenotypic spectrum - range of findings

Robin Sequence
- Micrognathia

Robin Sequence
- Airway obstruction 2ndary to glossoptosis

Robin sequence
- Wide u-shaped cleft

Van Der Woude Syndrome
- Lower lip pits
- hypodontia
- absent central and lateral incisors
- cleft lip +/- palate
- cleft palate