Genetics and the most prevalent Syndromes

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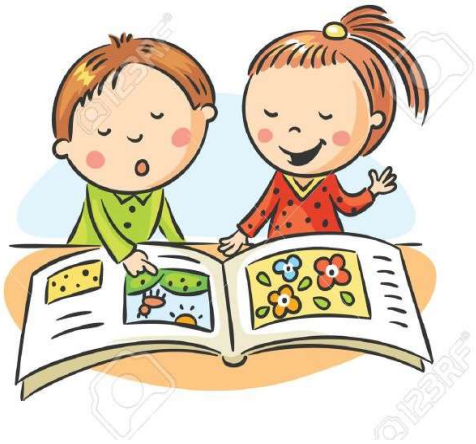
Goals

- The importance of Genetics
- Congenital malformation categories
- Approach to the dysmorphic child

The importance of Genetics

- ٢ to ٤% of neonate
- ٧% of children at the age of one year old
- ٣٠ to ٥٠% of the hospitalized children



Congenital malformation categories

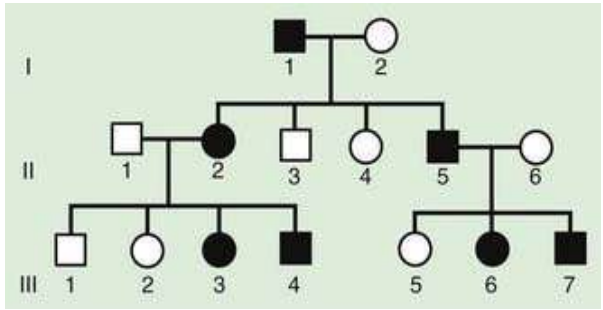
- Single gene mutation, ۶%
- Chromosomal disorders, ۷٫۵%
- Exposure to teratogen, ۶%
- Multifactorially inherited (polygenic), ۲۰%
- Unusual pattern of inheritance (mitochondrial, uniparental disomy, expansion of a trinucleotide repeat), ۲ to ۳%
- No category, most cases

Pedigree drawing

- Proband
- Affected individual
- Carriers
- At least three generation

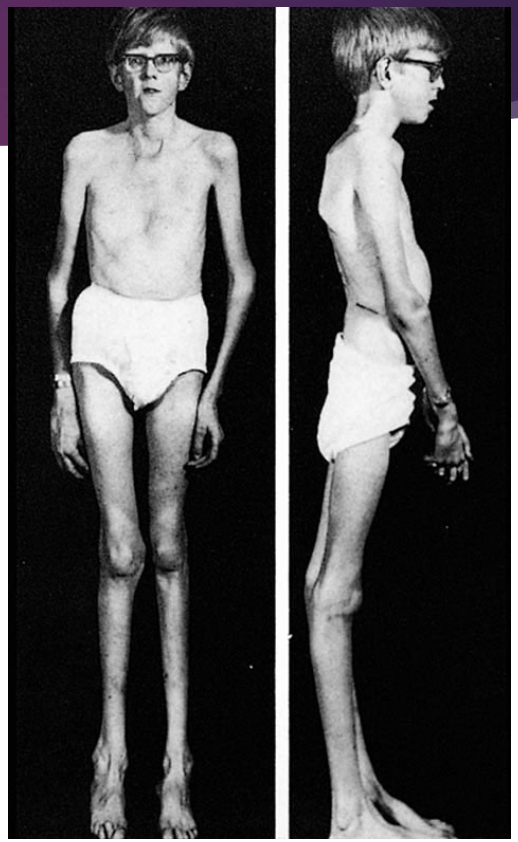
Autosomal dominant disorders

- Both male and female equally
- Each child has ٥٠% chance
- Every generation is affected
- Male to male is possible
- Examples: Achondroplasia, Marfan Syndrome, Neurofibromatosis ١ and ٢, Hereditary angioedema, Huntington Disease, Myotonic dystrophy



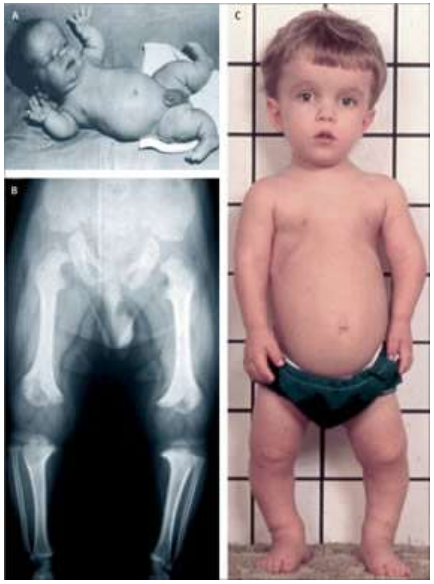
Marfan syndrome

- Long and narrow face
- Long legs, arms, fingers and toes
- Flexible joints
- Heart murmurs
- Dislocated lens



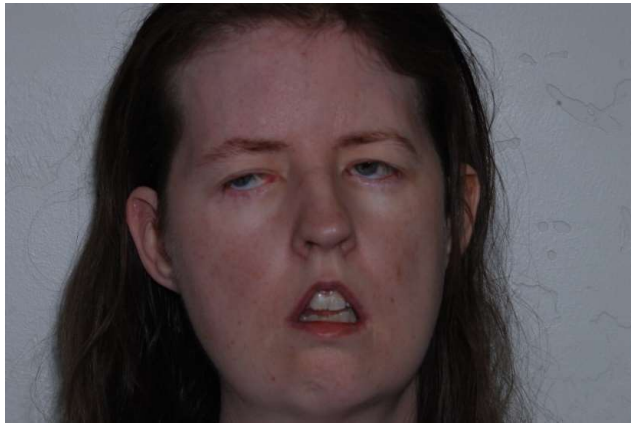
Achondroplasia

- Large head, prominent forehead, small midface
- Bowed legs, dwarfism, short arm and legs
- Small fingers, ring finger far apart from middle finger
- Spinal curve
- Apnea



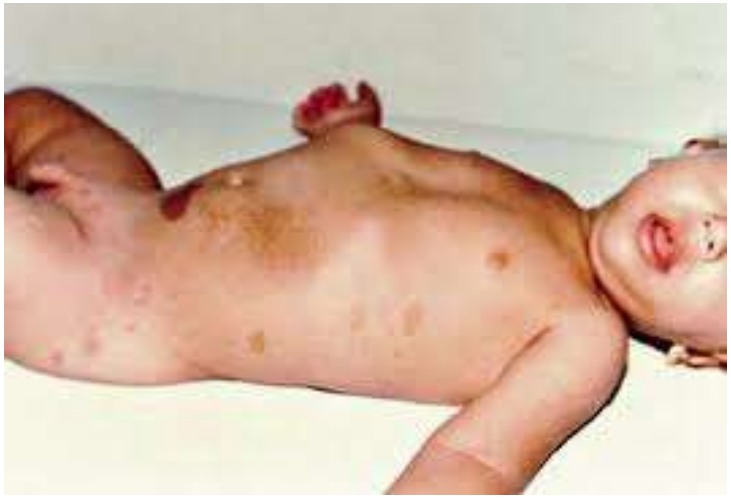
Myotonic dystrophy

- Decreased fetal movement, hypotonia
- Feeding difficulty, weak sucking, myotonia
- Cardiac problems
- Delayed motor and speech development
- Typical faces



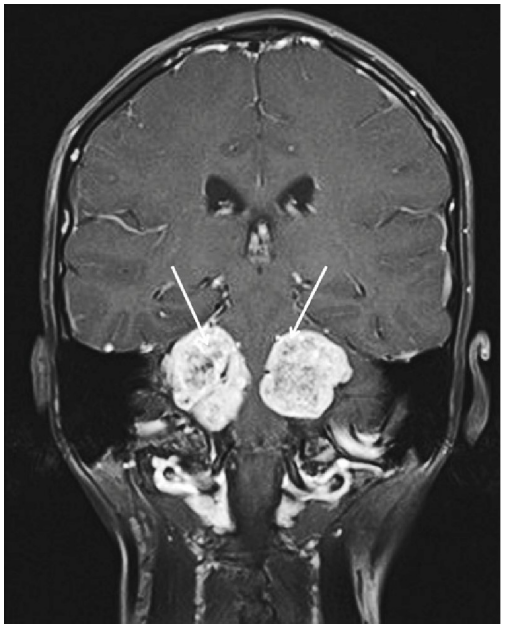
Neurofibromatosis ١

- Café au lait spots, neurofibroma
- Freckling of non sun exposed area
- Lisch nodules of iris
- Optic nerve glioma
- Tibia bowing, scoliosis

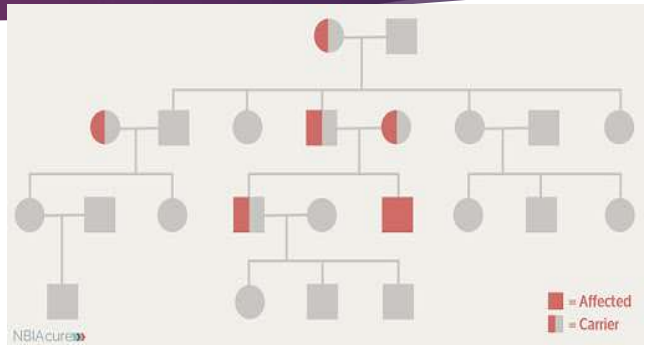


Neurofibromatosis ٢

- Bilateral vestibular nerve schwannomas
- Peripheral nerve tumors, neuropathy
- Cranial and spinal meningioma
- Early onset cataract



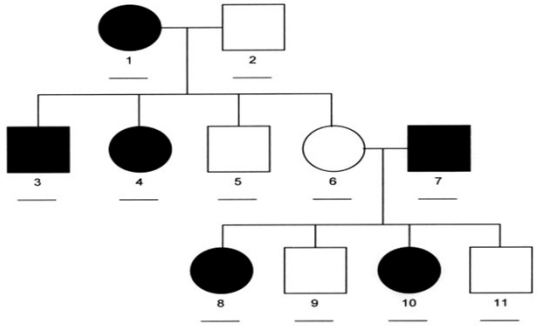
Autosomal recessive disorders



- Each sibling has ٢٥% chance
- Siblings are affected, not parents or offspring
- Normal siblings may be carrier
- Male and female equal
- Examples: Congenital adrenal hyperplasia, Sickle cell anemia, Cystic fibrosis, Phenylketonuria, Gaucher Disease, Friedreich ataxia

X linked dominant disorders

- Both male and female
- Female affects less severe



- Examples: X linked vitamin D resistant or hypophosphatemic rickets, Incontinentia pigmenti, Rett syndrome

Rett syndrome

- Normal at birth
- Developmental regression in the first year of life
- Handwashing posture
- Microcephaly
- Seizure
- Autism at the age of ٢