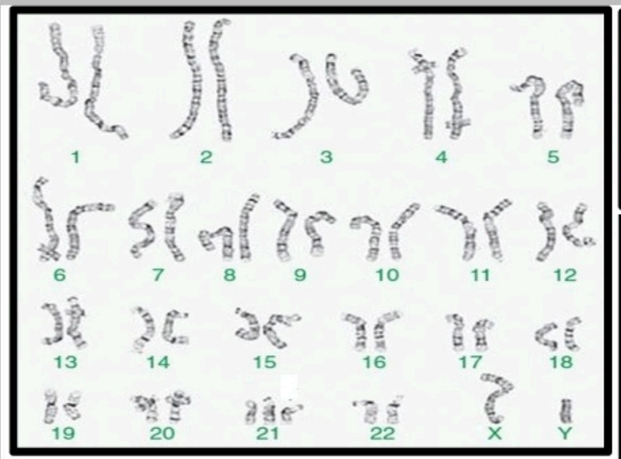
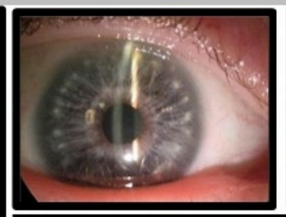
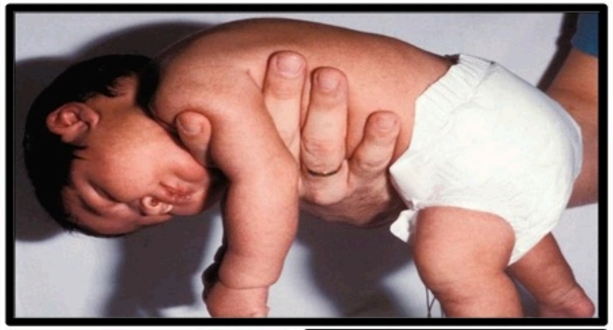
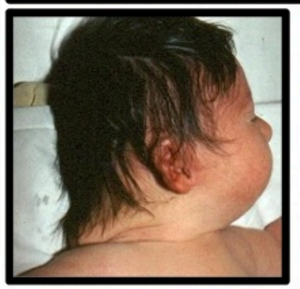
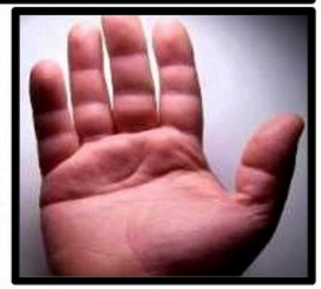


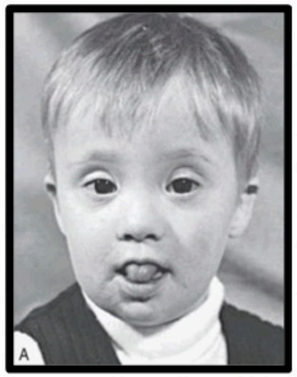


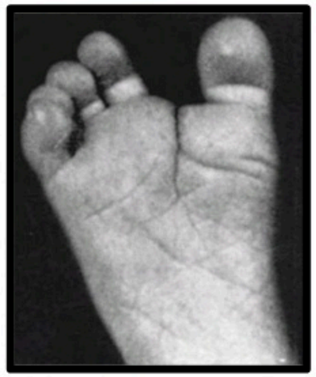
1) Trisomy 21 (Down syndrome)











❖ **Ophthalmology clinical features** :
- ✓ Myopia
- ✓ Cataract
- ✓ Glaucoma
- ✓ Brushfield spots (speckled irises)
- ✓ Nystagmus
- ✓ Strabismus

Screen at birth, 6 mo. Than annually

Pediatric On Squares

❖ **Musculoskeletal clinical features** :
- ✓ Short neck, redundant skin
- ✓ Short metacarpals and phalanges
- ✓ Short 5th digit with Clinodactyly
- ✓ Single transverse palmer creases
- ✓ Wide gap between 1st & 2nd toes
- ✓ Short sternum
- ✓ Atlantoaxial instability (screen each visit)
- ✓ Hip dysplasia
- ✓ Recurrent joints dislocation

❖ **Cardiac clinical finding**:
- ✓ Endocardial cushing defect (AVSD) **(MOST COMMON)**
- ✓ Ventricular septal defect
- ✓ Atrial septal defect
- ✓ Patent ductus arteriosus
- ✓ Pulmonary hypertension

Screen at birth

❖ **Facial clinical features** :
- ✓ Brachycephaly
- ✓ Up slanted palpebral fissures
- ✓ Epicanthal folds
- ✓ Three fontanels
- ✓ Flat nasal bridge , small nose
- ✓ Short hard palate
- ✓ Microcephaly
- ✓ Low set ears
- ✓ Hearing loss
- ✓ Recurrent otitis media

Hearing screen Screen at birth than annually

❖ **Hematological finding**:
- ✓ Transient myeloproliferative dis.
- ✓ ALL (> 1y.o)
- ✓ AML (< 1y.o)

Screen at birth

❖ **Endocrine finding**:
- ✓ **Hypothyroidism**
- ✓ Hyperthyroidism
- ✓ Diabetes mellitus
- ✓ Infertility
- ✓ Obesity

Screen at birth, 6 mo. than annually

❖ **Respiratory finding**:
- ✓ Obstructive sleep apnea
- ✓ Recurrent infections

Screen at 1 Y.O

❖ **Neuropsychiatric finding**:
- ✓ **Hypotonia**
- ✓ Developmental delay
- ✓ Autism
- ✓ Seizure

Screen each visit

❖ **Gastrointestinal finding**:
- ✓ Duodenal atresia
- ✓ Annular pancreas
- ✓ Tracheoesophageal fistula
- ✓ Hirschsprung disease
- ✓ Imperforated anus
- ✓ Neonatal cholestasis
- ✓ Celiac disease (Screen at 2y.o)

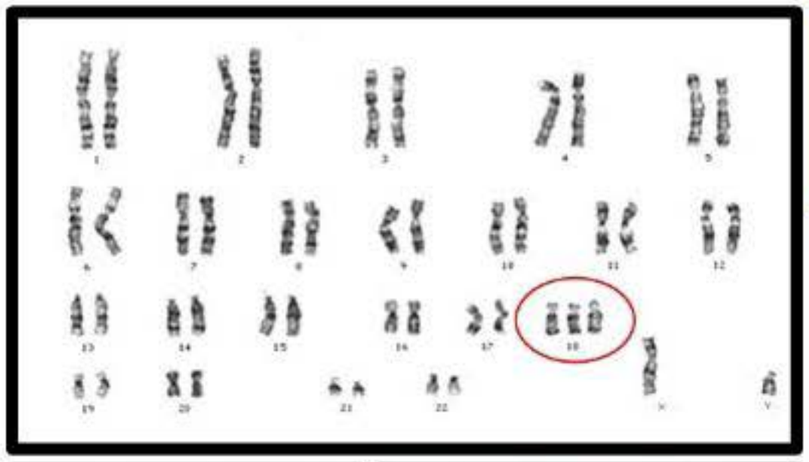
❖ **Causes:**
- ✓ Nondisjunction (95%)
- ✓ Translocation (4%)
- ✓ Mosaic (1%)

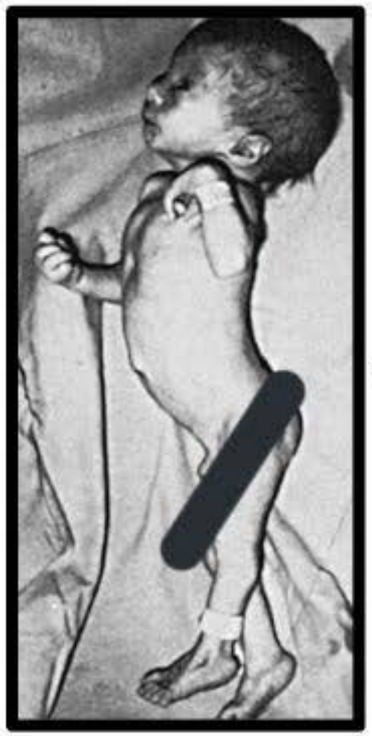
Recurrence 1%

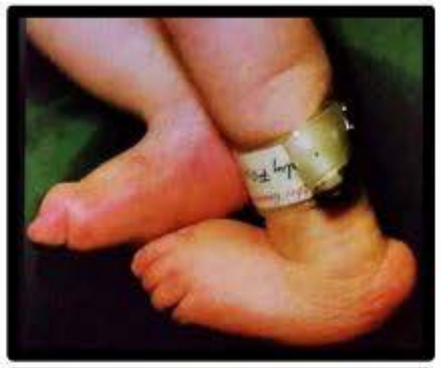
When the recurrence become 100%?
- ✓ If a parent has 21:21 translocation

❖ **Prenatal screen lab finding?**
- ✓ Low α feto protein
- ✓ Low estriol
- ✓ High hCG and high inhibin

MATERNAL AGE (YR)	DOWN SYNDROME
All ages	1 in 650
20	1:1500
30	1:1000
35	1:385
40	1:110
45	1:37







2) Trisomy 18 (Edwards syndrome)

- **Clinical features**
  - ✓ **Rocker bottom feet**
  - ✓ **Overlapping fingers**
  - ✓ **Clenched fist**
  - ✓ Short sternum
  - ✓ Hypoplastic nails
  - ✓ Hypoplastic nasal alae
  - ✓ IUGR
  - ✓ Intellectual disability
  - ✓ Microcephaly
  - ✓ Hypertonia
  - ✓ Prominent occiput
  - ✓ Micrognathia
  - ✓ Limited hip abduction
  - ✓ Cleft lip / palate

2nd most common autosomal trisomy

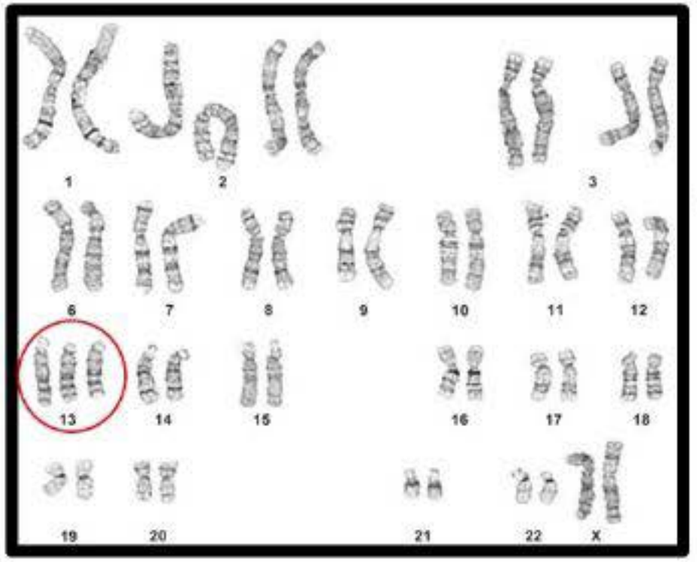
- **Most common cardiac defect ?**
  - ✓ **Ventricular septal defect (VSD)**
  - ✓ Atrial septal defect (ASD)
  - ✓ PDA

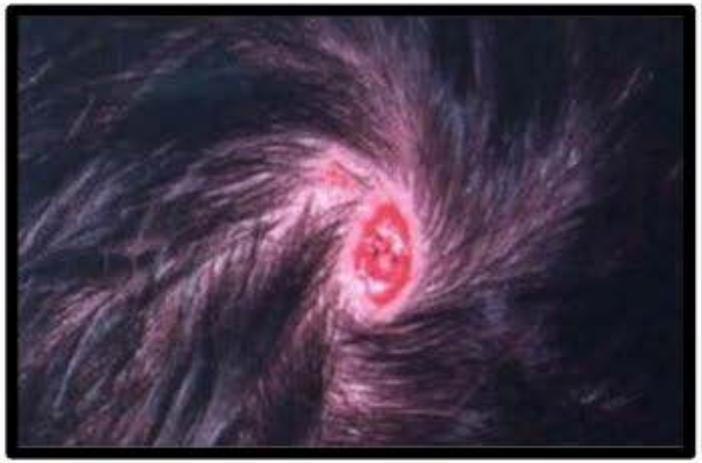
- **Most common cause of death?**
  - ✓ Central apnea

- **Prenatal screen lab finding?**
  - ✓ Low α feto protein
  - ✓ Low estriol
  - ✓ Low hCG

Pediatric On Squares

- **Prognosis**
  - ✓ 50% die in the 1st week of life
  - ✓ 90% die by 1 year of age





3) Trisomy 13 (Patau syndrome)

❖ Clinical features

- ✓ **Midline defect**
- ✓ **Aplasia cutis congenita**
- ✓ Microphthalmia
- ✓ Microcephaly
- ✓ Postaxial polydactyly
- ✓ Hypotonia
- ✓ Holoprosencephaly
- ✓ Hypoplastic / absent ribs
- ✓ Abdominal wall defect
- ✓ Deafness
- ✓ Colobomas
- ✓ Capillary hemangioma
- ✓ Genital anomalies
- ✓ Clenched fist

3rd most common autosomal trisomy

❖ Most common cardiac defect ?

1. **Ventricular septal defect (VSD)**
2. Atrial septal defect (ASD)
3. PDA

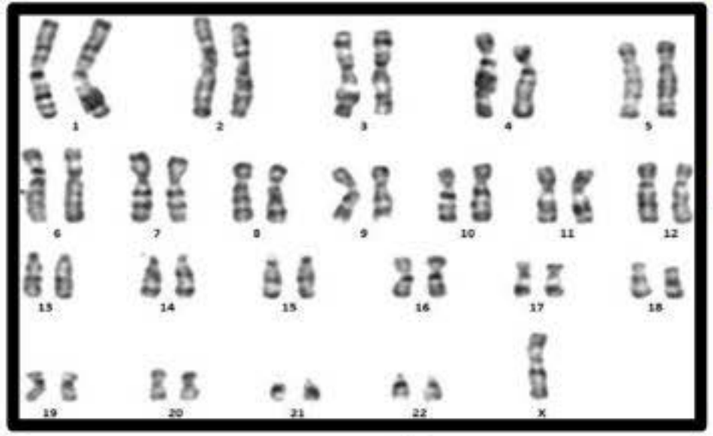
❖ Most common cause of death?

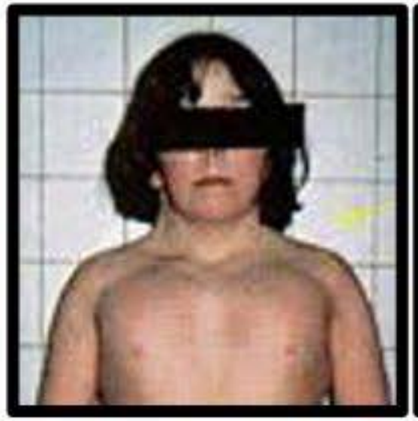
✓ Central apnea

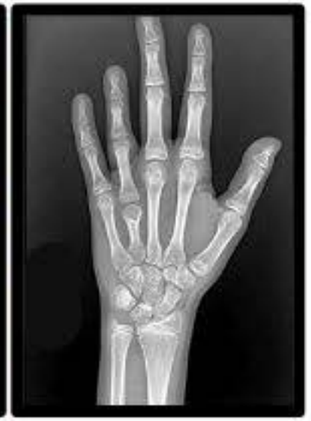
Pediatric On Squares

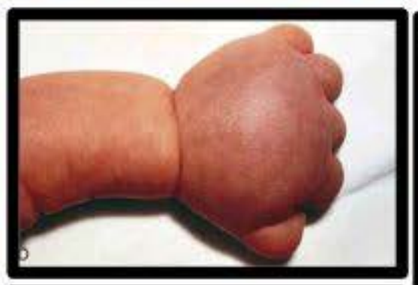
❖ Prognosis

- ✓ 70% die in the 1st 3 months of life
- ✓ 95% die by 3 years of age
- ✓ Rarely reach up to 10 years









4) Turner Syndrome

❖ Clinical features

- ✓ Short stature
- ✓ Lymphedema of hands & feet
- ✓ Shield chest
- ✓ Cubitus valgus
- ✓ Low posterior hairline
- ✓ Posterior rotated ears
- ✓ Webbed neck
- ✓ Short 4th metacarpal bone
- ✓ Normal intelligence (except Math)

❖ What is the most consistent finding?

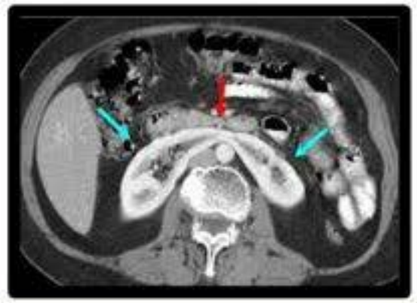
- ✓ Short stature (SHOX gene)
- ✓ Ovarian dysgenesis

❖ What is the most common cardiac defect?

1. Bicuspid aortic valve
2. Coarctation of aorta

❖ Labs finding ?

- ✓ High FSH
- ✓ High FSH/LH
- ✓ Low estradiol



❖ What is the most common renal anomaly ?

- ✓ Horseshoe kidney
- ✓ Double collecting system

❖ Increases risk of :

- ✓ Hypothyroidism
- ✓ Hashimoto disease
- ✓ Celiac disease
- ✓ Inflammatory bowel disease

Pediatric On Squares

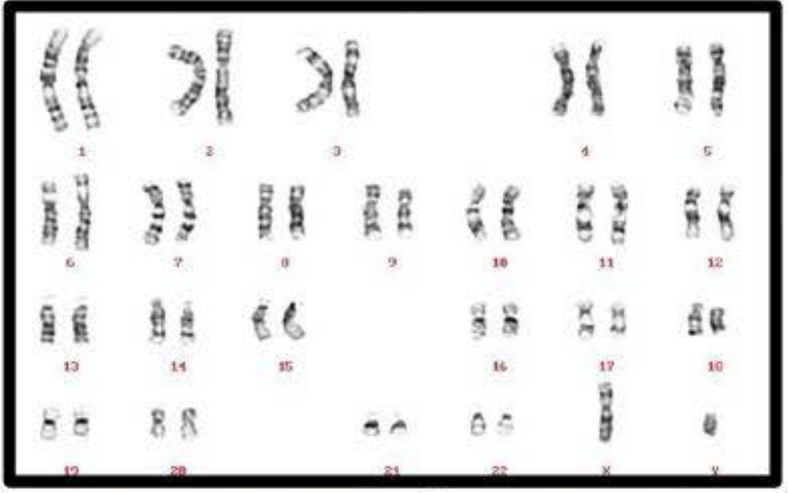
❖ Increases risk of which Malignancy ?

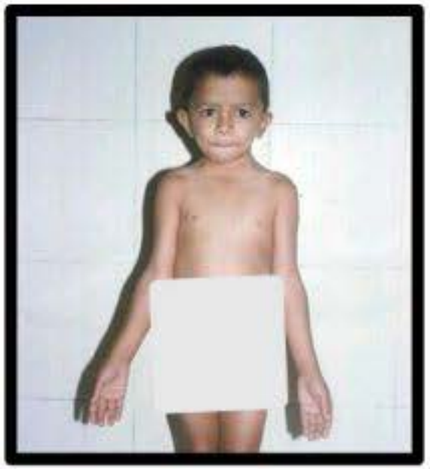
- ✓ Gonadoblastoma

❖ Treatment

- ✓ Growth hormone
- ✓ Estrogen therapy

5) Noonan Syndrome





❖ Clinical features

- ✓ **Down slanting palpebral fissures**
- ✓ Short stature
- ✓ Shield chest
- ✓ Cubitus valgus
- ✓ Low posterior hairline
- ✓ Webbed neck
- ✓ Posterior rotated ears
- ✓ Cryptorchidism
- ✓ Intellectual disability
- ✓ Delayed puberty
- ✓ Ptosis

Pediatric On Squares

❖ Mode of inheritance?

- ✓ Autosomal dominant
- ✓ PTPN11 gene on 12q24
- ✓ Normal karyotype

❖ What is the most common cardiac defect?

1. Pulmonary stenosis
2. Hypertrophic cardiomyopathy

❖ Labs finding ?

- ✓ Clotting factor deficiencies
  - Mainly factor XI & XII
- ✓ Abnormal platelet count/function

❖ Increases risk of which Malignancy ?

- ✓ Acute lymphocytic leukemia (ALL)
- ✓ Chronic myeloid leukemia (CML)

❖ Treatment

- ✓ Growth hormone

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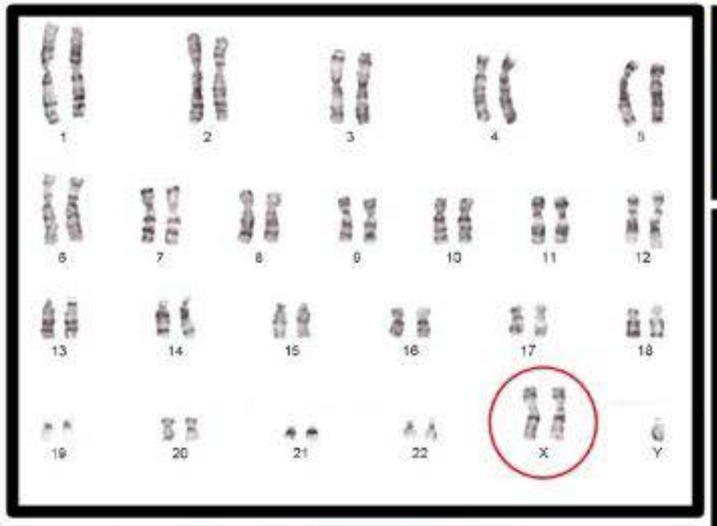

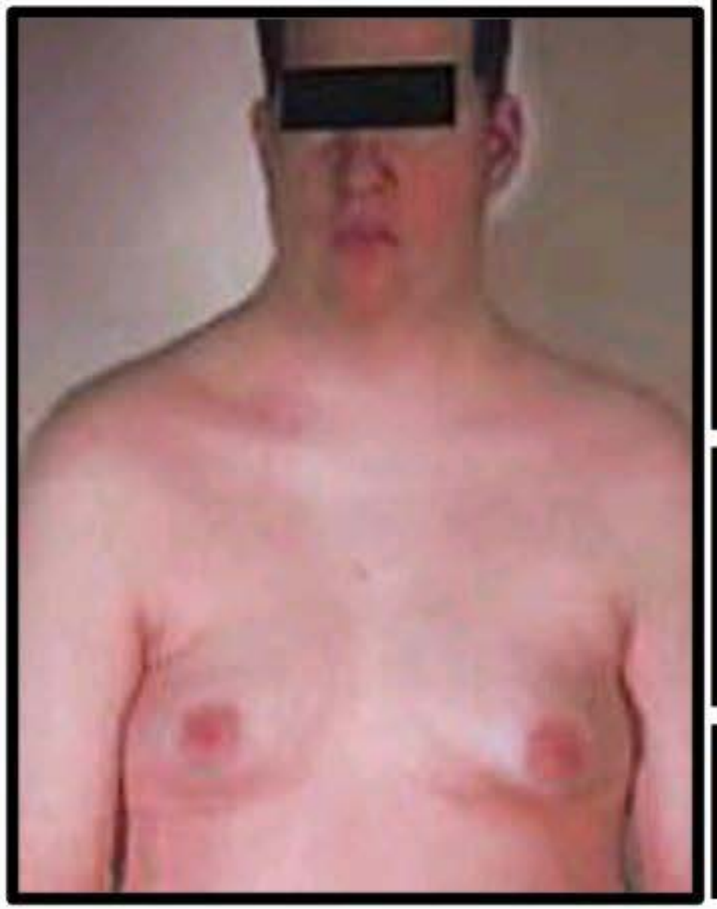

6) Klinefelter Syndrome XXY

❖ Clinical features

- ✓ **Gynecomastia**
- ✓ Tall stature
- ✓ **Small testes**
- ✓ Micropenis
- ✓ Normal pubic and axillary hair
- ✓ Intellectual disability
- ✓ Infertility
- ✓ Behavioral problems

Pediatric On Squores

The most common cause of primary hypogonadism and infertility in males

Recurrence rate 1 – 2 %

❖ Laboratory finding ?

- ✓ Low testosterone
- ✓ High FSH/LH

❖ Increases risk of :

- ✓ Pulmonary disease
- ✓ Varicose vain
- ✓ ADHD

❖ Increases risk of which Malignancies ?

- ✓ Breast cancer
- ✓ Testicular cancer

❖ What is the most effective treatment ?

- ✓ Testosterone replacement therapy

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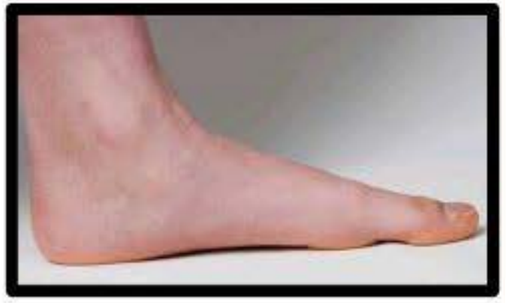


7) Fragile X Syndrome

❖ Clinical features

- ✓ Long narrow face
- ✓ Prominent jaw
- ✓ Big ears
- ✓ Large hand and feet
- ✓ **Macroorchidism**
- ✓ Hyperextensible joints
- ✓ Intellectual disability
- ✓ Behavioral problems (Autism, ADHD)
- ✓ Speech delay
- ✓ Pes planus (Flat foot)

Pediatric On Squares
@OnSquares

❖ Mode of inheritance?

- ✓ X-linked dominant inheritance
- ✓ > 200 **CGG** repeats in FMR1 gene

❖ What is the most common cardiac defect?

- ✓ **Mitral valve prolapse**

The most common form of **inherited** intellectual disability

But

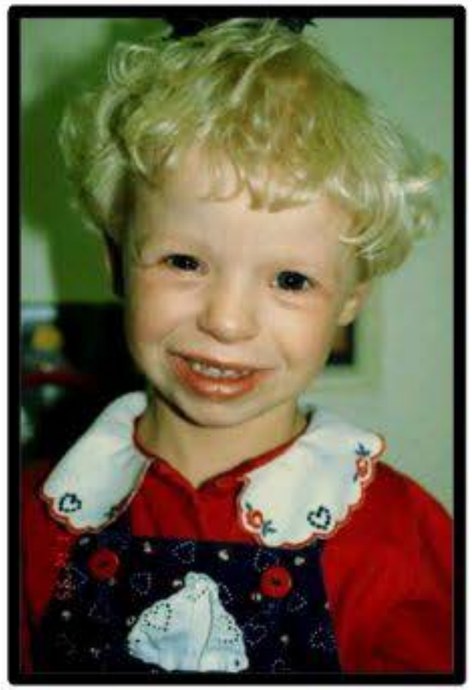
The 2nd most common **genetic** intellectual disability (after trisomy 21)

❖ How to confirm the diagnosis ?

- ✓ Molecular genetic test for CGG allele repeat size

❖ Can a female have fragile X syndrome?

- ✓ Yes, but milder symptoms
- ✓ Premature ovarian failure



8) William Syndrome

❖ **Mode of inheritance?**
- ✓ Autosomal dominant
- ✓ Microdeletion 7q

❖ **Laboratory finding ?**
- ✓ Hypercalcemia

❖ **Most common cardiac defect ?**
- ✓ **Supravalvular aortic stenosis (80%)**
- ✓ Peripheral pulmonic stenosis
- ✓ Pulmonary valvular stenosis

❖ **Clinical features**
- ✓ Long philtrum (**Elfin face**)
- ✓ Broad mouth.
- ✓ Short nose with bulbous nasal tip.
- ✓ **Cocktail personality**
- ✓ Mental retardation
- ✓ Joint laxity

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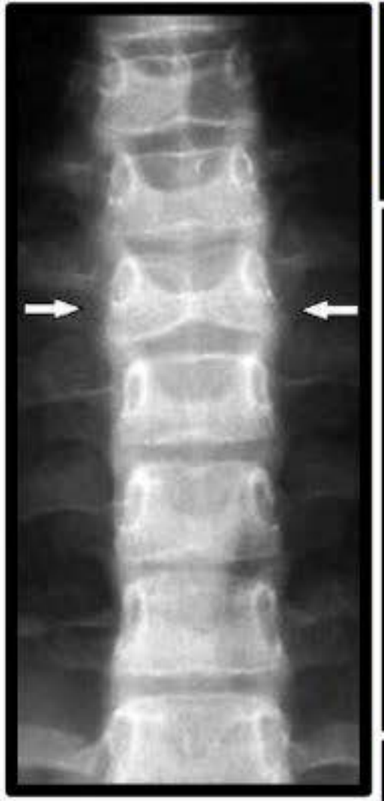
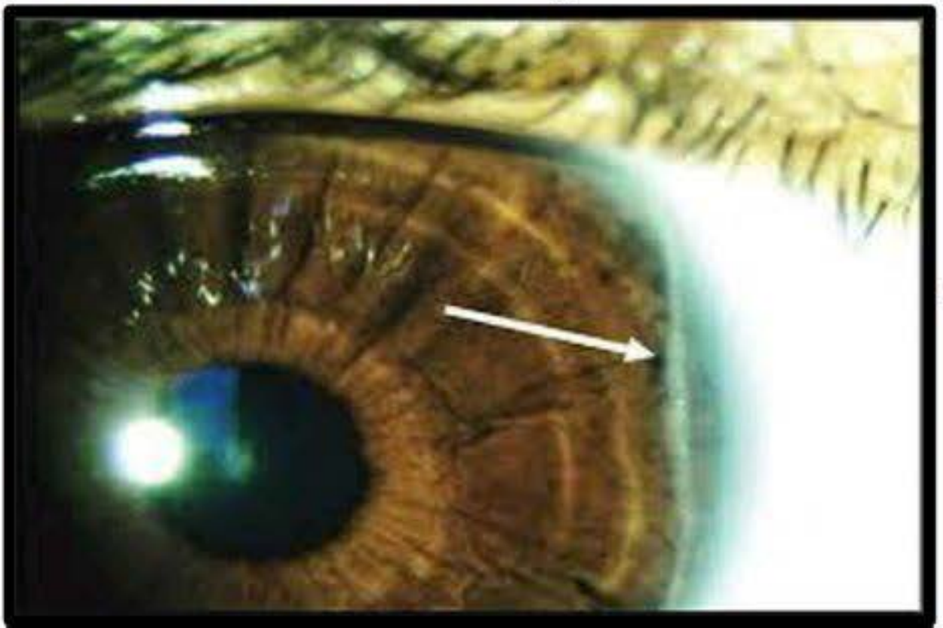
❖ **Eye problems ?**
- ✓ Stellate irises
- ✓ Strabismus

Pediatric On Squares

❖ **The AAP recommends annual cardiology evaluations for patients with Williams syndrome.**





9) Alagille syndrome

❖ Clinical features

- ✓ **Triangular face with pointed chin**
- ✓ Long nose
- ✓ Cholestatic jaundice
- ✓ **Posterior embryotoxon**
- ✓ **Butterfly vertebrae**

Liver biopsy show

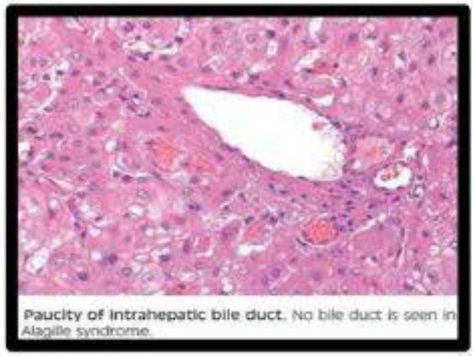
↓

Paucity of bile duct

❖ Mode of inheritance?

- ✓ Autosomal dominant
- ✓ 20p12 deletion
- ✓ JAG1 gene

Pediatric On Squares
@OnSquares



Paucity of intrahepatic bile duct. No bile duct is seen in Alagille syndrome.

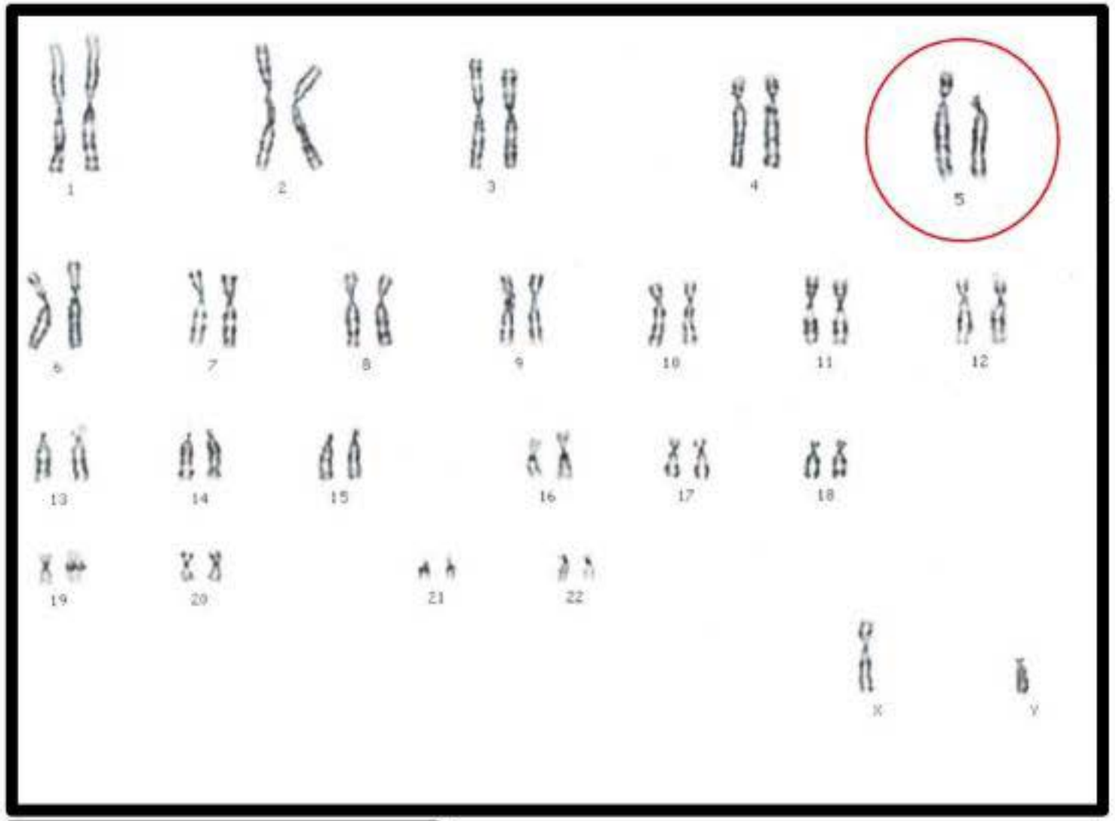
❖ Most common cardiac defect ?

❖ Peripheral pulmonary artery stenosis / pulmonary valve stenosis

❖ Treatment ?

- ✓ Ursodeoxycholic acid (UDCA)
- ✓ Liver transplant

10) Cri du chat syndrome



❖ Clinical features

- ✓ **High pitched (Cat's like) cry** (larynx anatomical changes)
- ✓ Moon face with wide spaced eyes
- ✓ Wide and flat nasal bridge
- ✓ Hypotonia
- ✓ Down slanting palpebral fissures
- ✓ Microcephaly
- ✓ Feeding difficulties
- ✓ Intellectual disabilities
- ✓ Failure to thrive

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❖ Mode of inheritance?

- ✓ De novo mutation
- ✓ 5p deletion

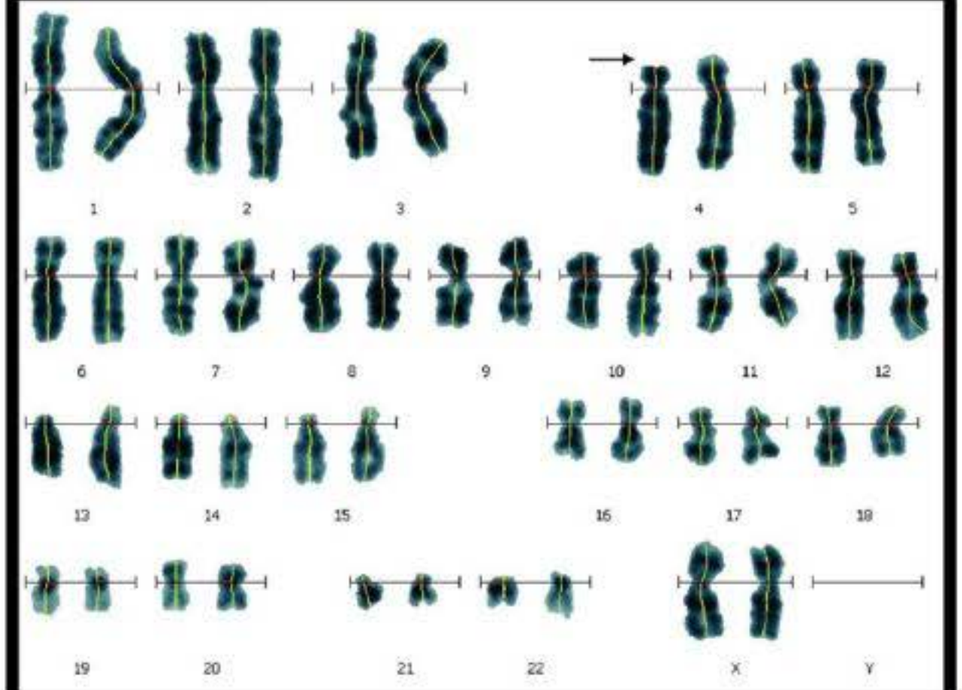
❖ Common cardiac defect ?

- ✓ **VSD, ASD, PDA & TOF**

Pediatric On Squares

11) Wolf–Hirschhorn syndrome

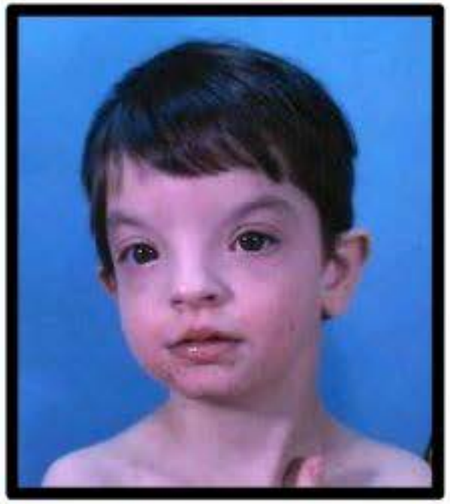
≠



❖ Clinical features:

- ✓ Microcephaly
- ✓ Hypertelorism
- ✓ Prominent glabella
- ✓ Frontal bossing
- ✓ **Greek helmet face**
- ✓ Beaked nose
- ✓ Hypotonia
- ✓ Seizures
- ✓ Cardiac defect

Pediatric On Squares



❖ Mode of inheritance?

- ✓ De novo
- ✓ Deletion of the short arm of chromosome 4

❖ Laboratory finding ?

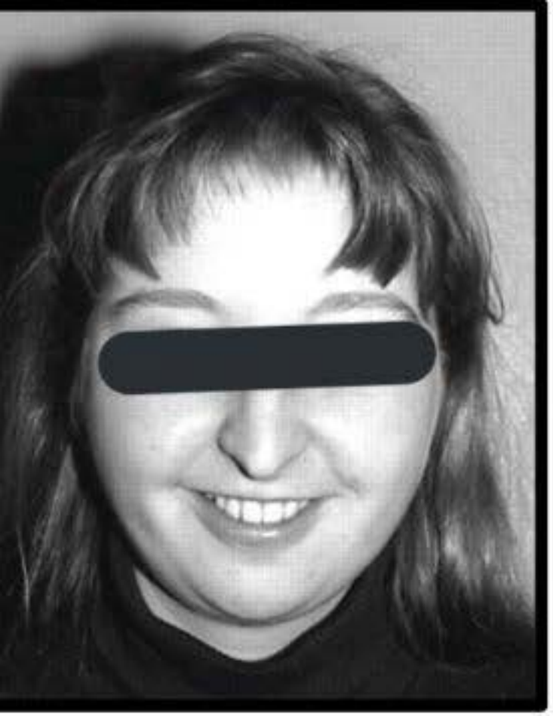
- ✓ IgA deficiency

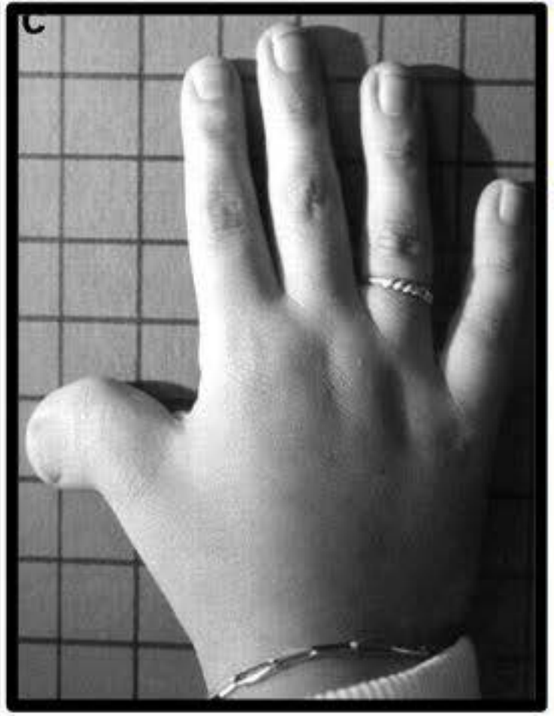
Wolf–Farm syndrome

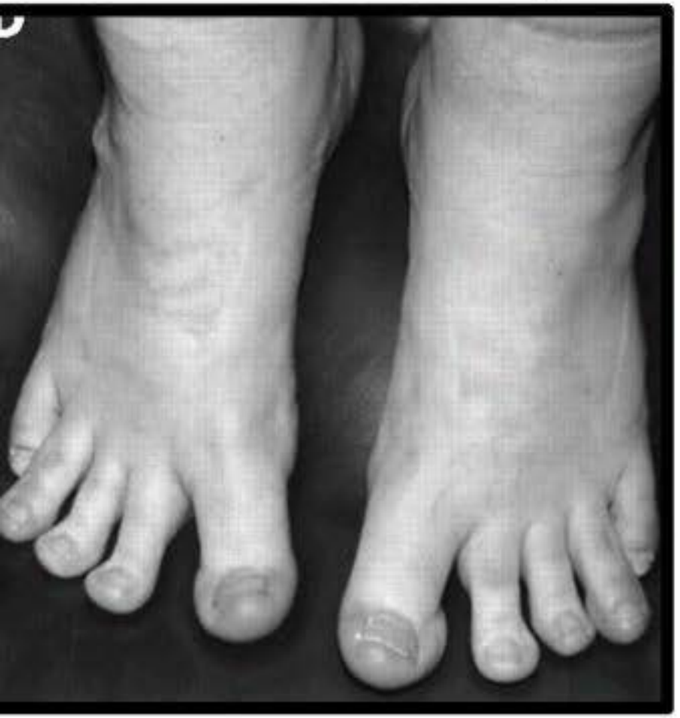
❖ Clinical features:

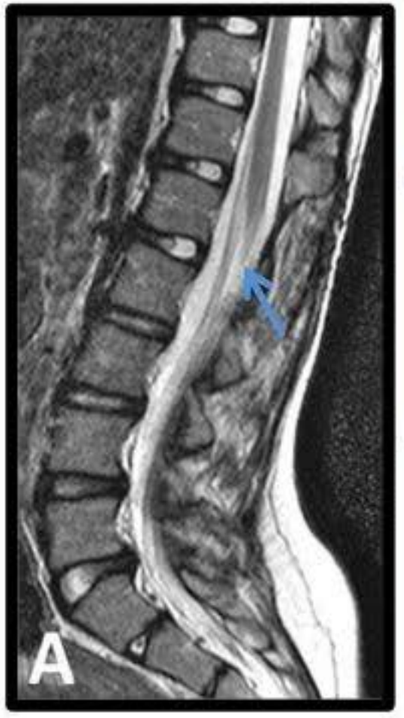
- ✓ Diabetes insipidus
- ✓ Diabetes mellitus
- ✓ Blindness
- ✓ Deafness

@OnSquares









12) Rubinstein Taybi Syndrome

❖ **Clinical features**

- ✓ **Broad thumbs and great toes**
- ✓ **Prominent nasal septum , beaked nose**
- ✓ Microcephaly
- ✓ Large nails
- ✓ Long eyelashes
- ✓ Spinal cord tethering
- ✓ Cardiac defect
- ✓ Growth & mental retardation

Pediatric On Squares
@OnSquares

❖ **Mode of inheritance?**

- ✓ Autosomal dominant
- ✓ Microdeletion 16q

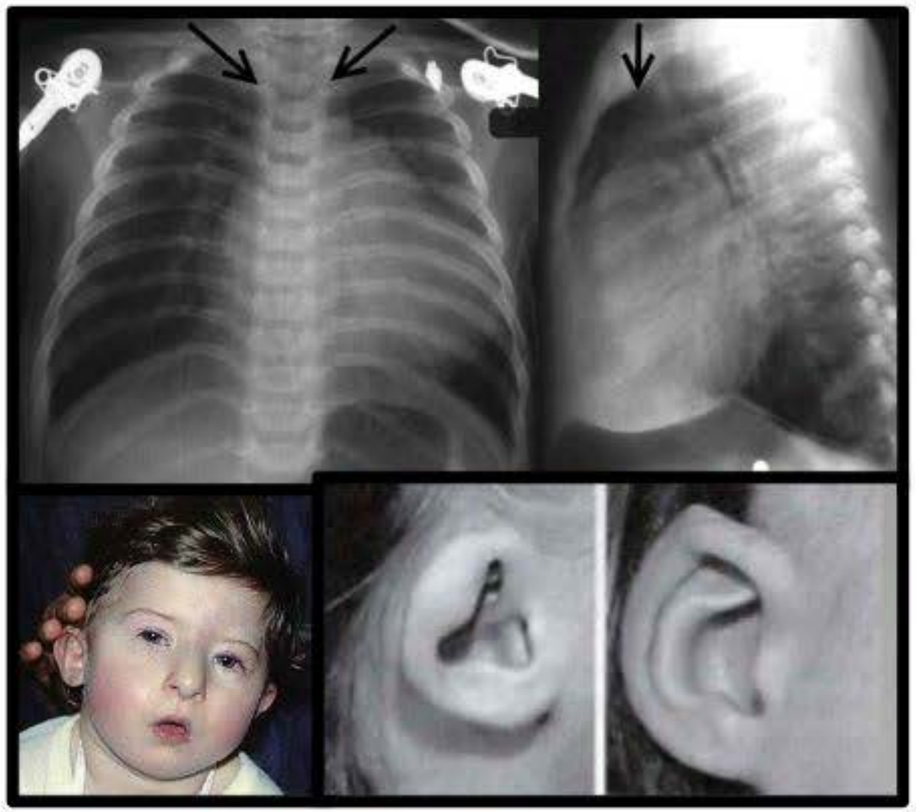
❖ **What is the most common cardiac defect?**

- ✓ Ventricular septal defect (VSD)

⚠ **CAUTION !!**

Rubinstein taybi syndrome have an increase risk of complications from anesthesia

13) DiGeorge syndrome (Velocardiofacial syndrome)



Clinical features (CATCH 22)

- ✓ Cardiac defect
- ✓ Abnormal face (Hooded eyelids, ptosis, hypoplasia of auricles)
- ✓ Thymic hypoplasia (T cell abnormalities)
- ✓ Cleft palate
- ✓ Parathyroid aplasia / hypoplasia
- ✓ Hypernasal speech (Velopharyngeal incompetence)
- ✓ Hypotonia in infancy
- ✓ Learning disabilities & behavioral problems

❖ **Other names ?**
- ✓ 22q11.2 deletion syndrom
- ✓ Shprintzen syndrome

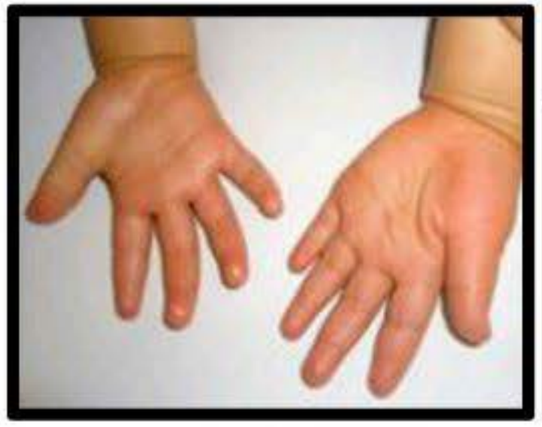
❖ **Mode of inheritance?**
- ✓ Autosomal dominant

❖ **Common cardiac defect?**
- ✓ Interrupted aortic arch
- ✓ Truncus arteriosus
- ✓ Tetralogy of Fallot
- ✓ VSD

❖ **Laboratory finding ?**
- ✓ Hypocalcemia
- ✓ Low PTH
- ✓ Low IgG

❖ **It is the MOST COMMON :**
- ✓ Microdeletion syndrome
- ✓ T-Cell disorder

❖ **Which vaccine should be avoided ?**
- ✓ live attenuated vaccine if immunodeficient



14) Angelman syndrome

- **Clinical features:**
  - ✓ Jerky ataxic movement "**Happy puppet**"
  - ✓ Inappropriate bouts of laughter
  - ✓ Excitable with hand flapping movement
  - ✓ Hypotonia
  - ✓ Fair hair
  - ✓ Seizure
  - ✓ Microcephaly
  - ✓ Severe Intellectual disabilities
  - ✓ Speech delay
  - ✓ Abnormal creases
  - ✓ Small and separated teeth

Pediatric On Squares
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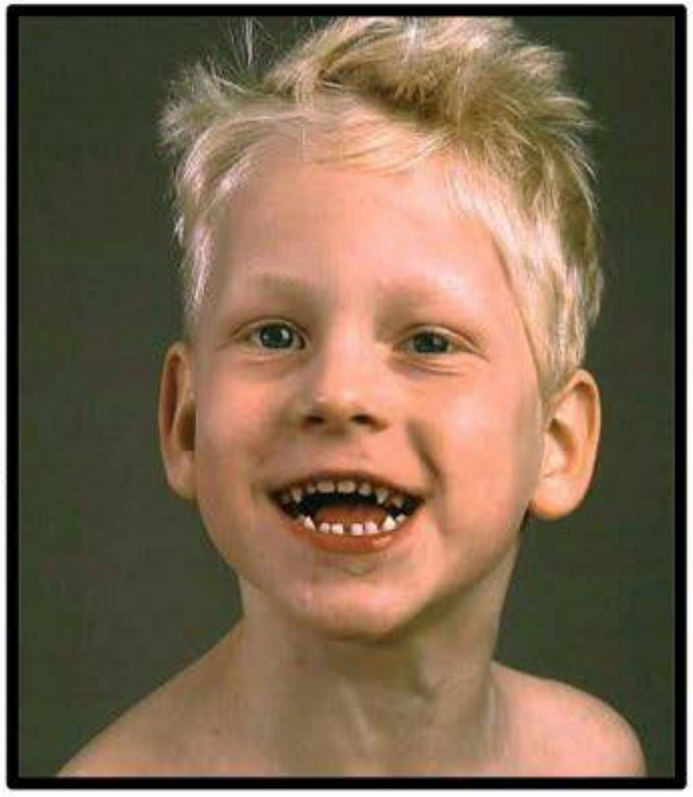
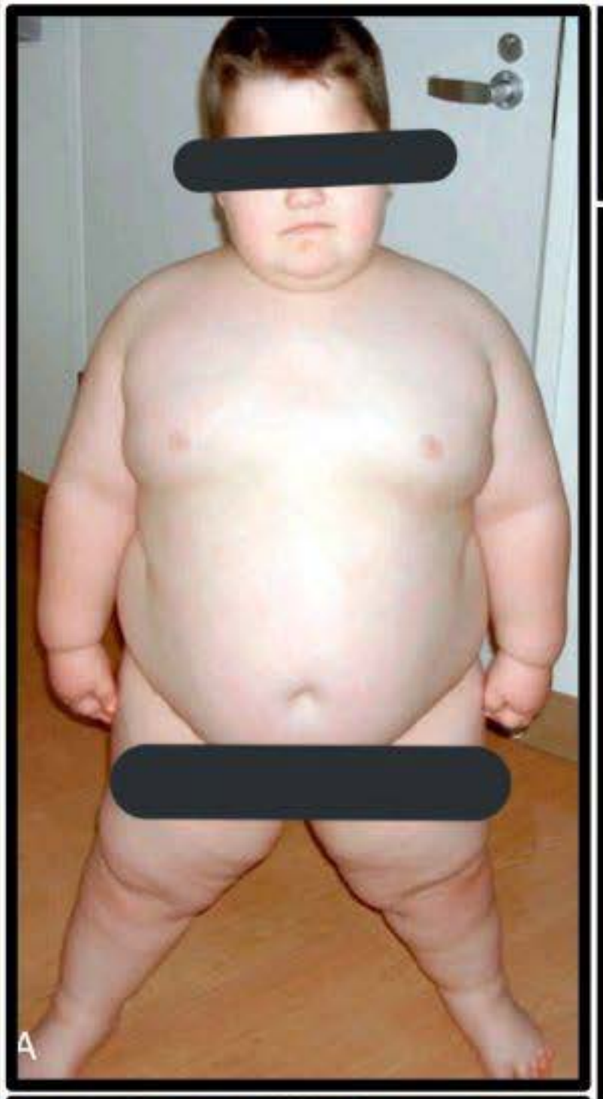
- **Mode of inheritance?**
  - ✓ Sporadic
  - ✓ Maternal derived deletion 15q11–13
  - ✓ Maternal UPD

- **Which medication should be avoided in seizure treatment?**
  - ✓ Carbamazepine
  - ✓ Vigabatrin





15) Prader Willi Syndrome (PWS)

❖ Clinical features in infancy

- ✓ Hypotonia
- ✓ feeding problems

❖ Clinical features in childhood

- ✓ Hyperphagia
- ✓ Central obesity
- ✓ Hypogonadism
- ✓ Short stature
- ✓ Small hand and feet
- ✓ Mild intellectual disability
- ✓ Almond-shaped eyes
- ✓ Behavioral disorder

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❖ Mode of inheritance?

- ✓ Sporadic
- ✓ Paternal derived deletion 15q11–13 (80%)
- ✓ Maternal UPD (20%)

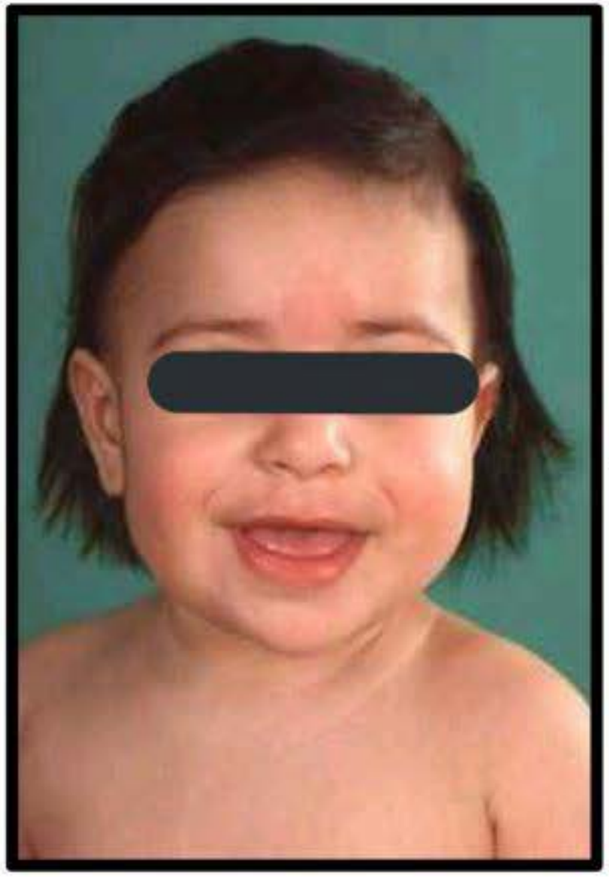
It is the most common syndromic form of obesity

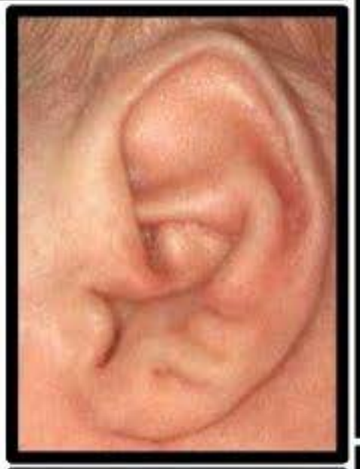
❖ Which medication had FDA approval to be used with PWS?

- ✓ Growth hormone therapy

❖ Why they develop short stature, central obesity & hypogonadism ?

- ✓ Secondary to Hypothalamic and pituitary dysfunction





16) Beckwith-Wiedemann syndrome

❖ Clinical features

- ✓ Large for gestational age
- ✓ Macroglossia
- ✓ Omphalocele
- ✓ **Ear lobe creases**
- ✓ **Posterior auricular pits**
- ✓ Hemihypertrophy
- ✓ Cryptorchidism
- ✓ Umbilical hernia
- ✓ Normal intelligence

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❖ Laboratory finding ?

- ✓ Hypoglycemia
- ✓ Hyperinsulinism
  - ➢ Due pancreatic islet hyperplasia

❖ Mode of inheritance?

- ✓ Sporadic
- ✓ Autosomal dominant
- ✓ 15q11 deletion

❖ Increases risk of which Malignancies ?

- ✓ Wilms tumor
- ✓ Hepatoblastoma

❖ Clinic follow up ?

- ✓ **Abdominal US**
  - Q3 months until 4 Y.O
- ✓ **Alpha fetoprotein**
  - Q3 months until 4 Y.O
- ✓ **Renal US**
  - Q3 months from 4-7 Y.O

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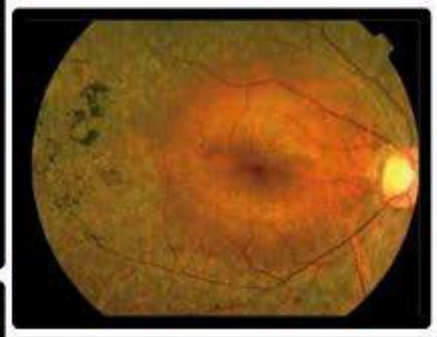
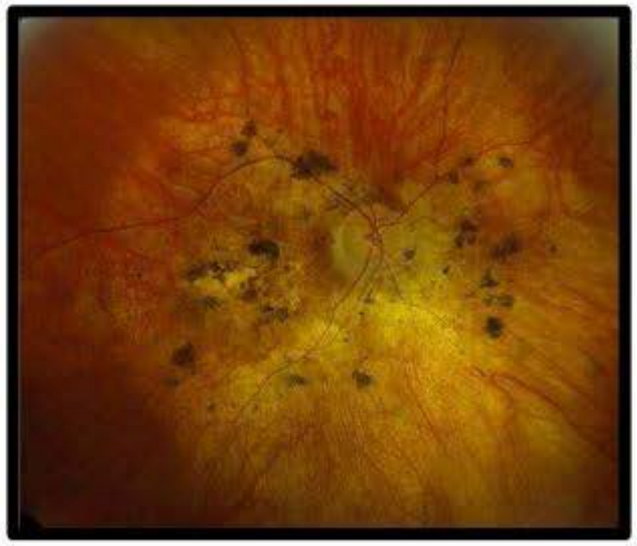
17) Bardet–Biedl syndrome (Laurence-Moon-Biedl syndrome)



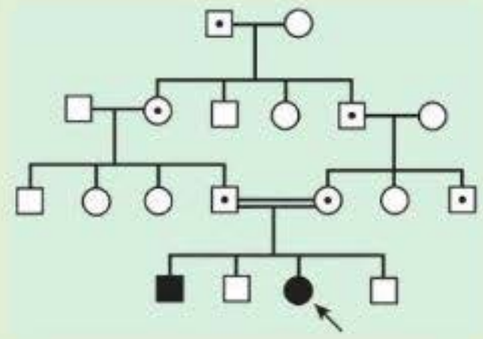


❖ Clinical features

- ✓ Obesity
- ✓ Postaxial polydactyly
- ✓ Hypogonadism
- ✓ Retinitis pigmentosa
- ✓ Rod-cone dystrophy
- ✓ Mental retardation
- ✓ Polyuria and polydipsia
- ✓ Renal anomalies

❖ Mode of inheritance?

- ✓ Autosomal recessive



❖ Most common gastrointestinal association?

- ✓ Hirschsprung disease

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❖ What is the cause of polyuria and polydipsia?

- ✓ Nephrogenic diabetes insipidus

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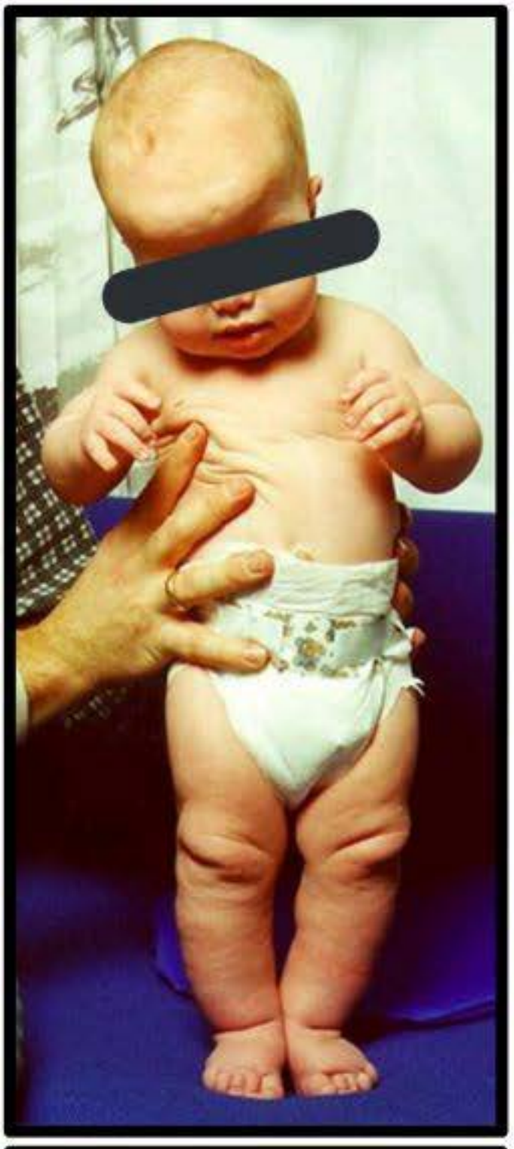

18) Sotos syndrome (cerebral gigantism)

❖Clinical features

- ✓ Large for gestational age
- ✓ Excessive growth in 1st year of life
- ✓ Large head , frontal prominence
- ✓ **Large hand and feet**
- ✓ **Premature tooth eruption**
- ✓ Intellectual disability
- ✓ Behavioral problems (Autism)
- ✓ Seizure
- ✓ Hypotonia
- ✓ Monotone voice

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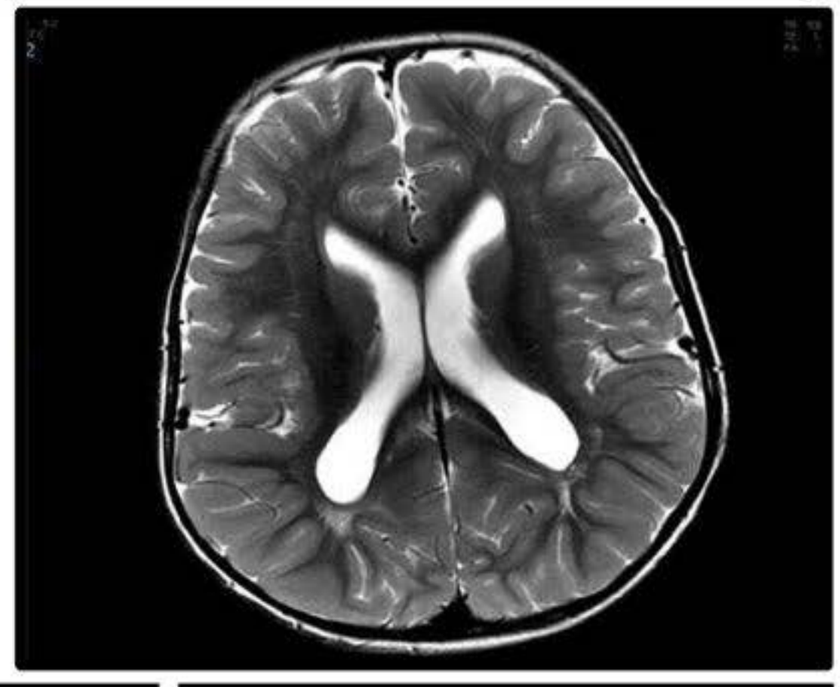
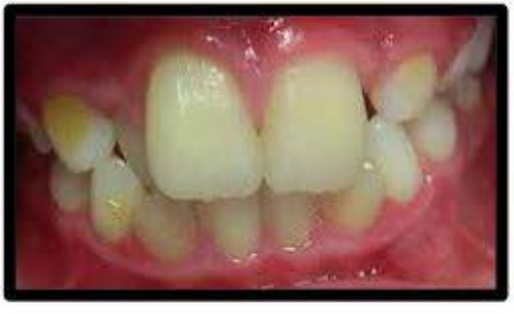
❖Radiological finding ?

✓**Ventriculomegaly**





❖ Mode of inheritance?

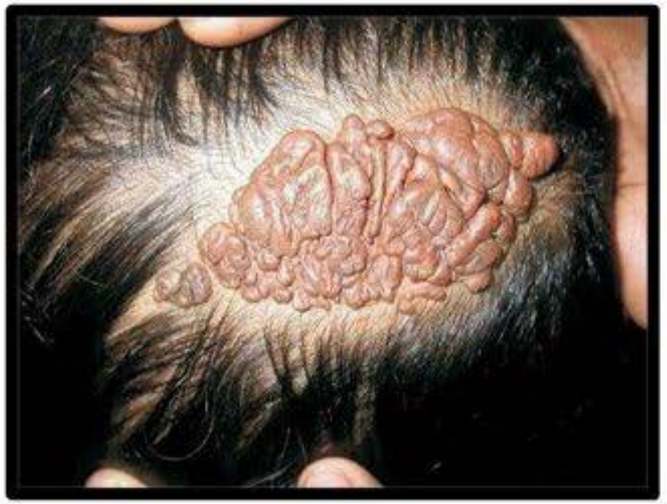
✓ Autosomal dominant

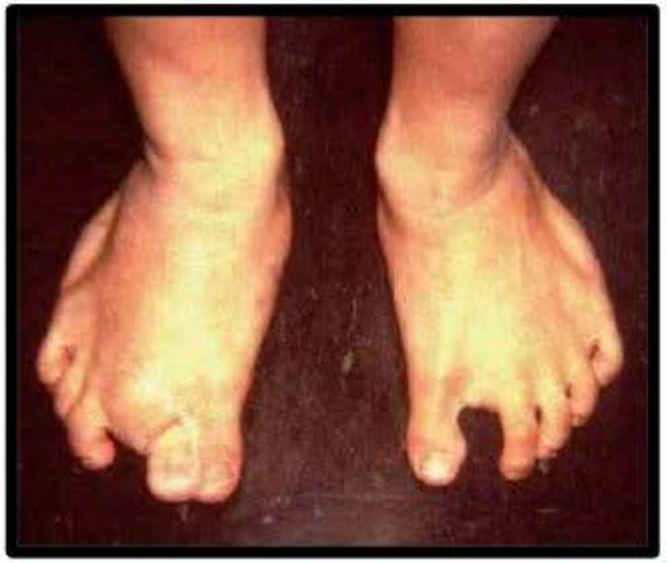
❖ Increases risk of which Malignancies ?

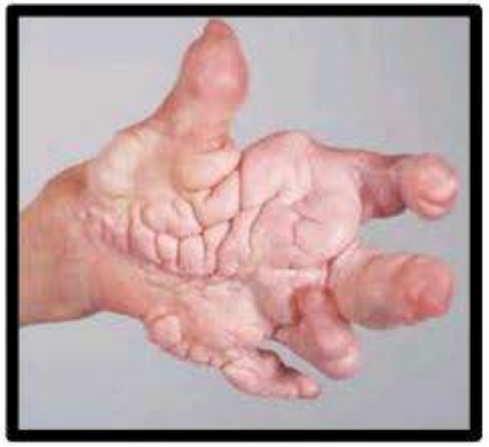
- ✓ Wilms tumor
- ✓ Hepatic carcinoma

❖ Prognosis

- ✓ Normal life expectancy
- ✓ Developmental delay







19) Proteus Syndrome

❖ **Clinical features**

- ✓ Macrodactyly
- ✓ Hemihypertrophy
- ✓ Skin changes (lipomas, vascular malformations, connective tissue nevi, epidermal)
- ✓ Scoliosis
- ✓ Tissue hypertrophy
- ✓ Accelerated growth
- ✓ Developmental delay
- ✓ Increase risk of malignancies

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❖ **What is Pathagnomic feature of Proteus syndrome ?**

- ✓ Connective tissue nevi, mainly "Cerebriform nevi"

❖ **Mode of inheritance?**

- ✓ Sporadic

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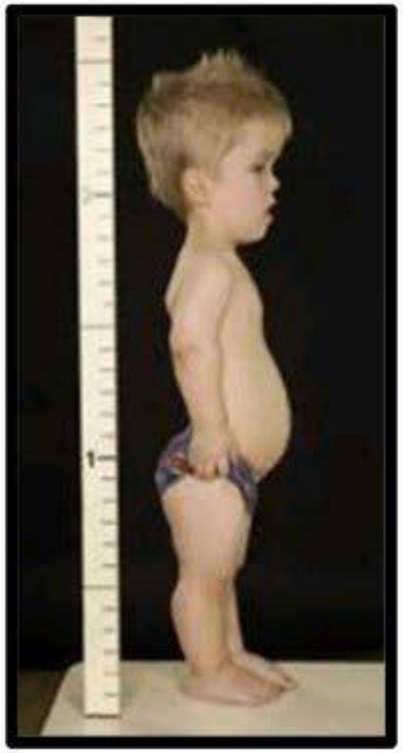
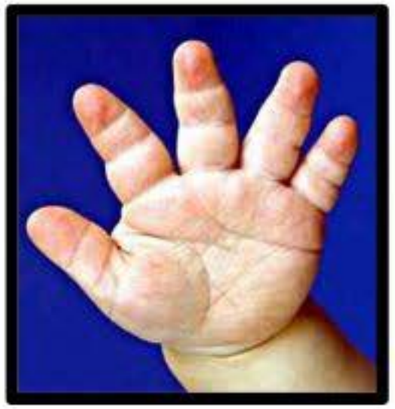
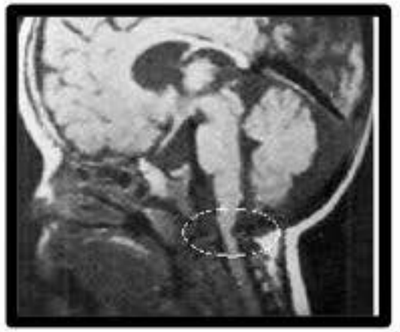
❖ **Increases risk of which Malignancies** ?

- ✓ Embryonic tumor

20) Achondroplasia







Clinical features

- Short stature below 3rd percentile.
- Rhizomelic Shorting (Short proximal long bones)
- Trident hands.
- Stenosis of foramen magnum.
- Macrocephaly.
- Midface hypoplasia
- Ice cream scoop shape femoral head.
- Squared-off iliac wings (Champagne glass sign).
- Increase risk of OSA & UAO.
- Delay in gross motor development.

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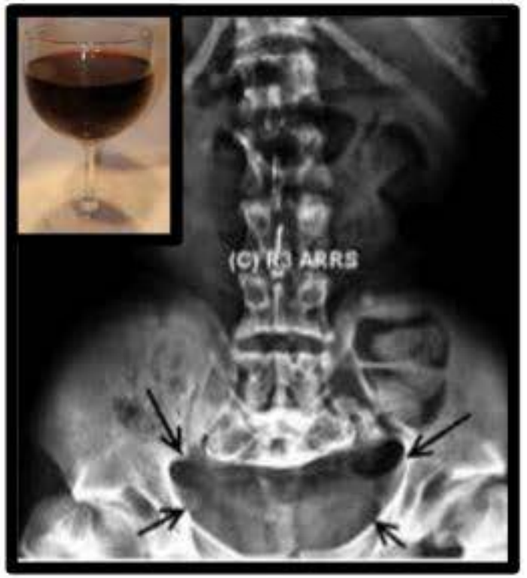


❖ Mode of inheritance?

- ✓ Autosomal Dominant
- ✓ Defect in FGFR-3

❖ Clinic follow up ?

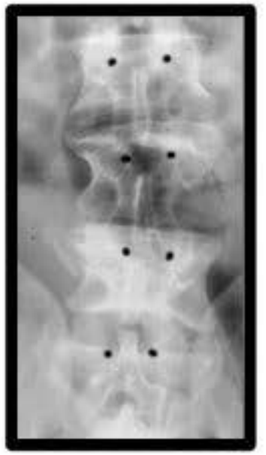
- ✓ Head circumference.
- ✓ Hearing test annually.
- ✓ Polysomnography.

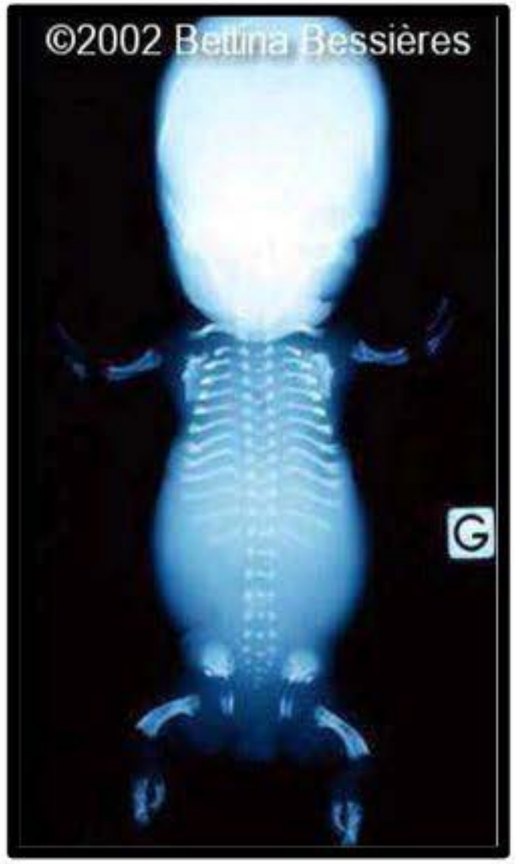
❖ Most common cause of death?

- ✓ Craniocervical junction compression

❖ Spinal X-ray specific finding ?

- ✓ Narrowed interpeduncular distances





21) Thanatophoric Dysplasia

Clinical features

- Rhizomelic Shorting
- Small chest
- Absent sciatic notch.
- Platyspondyly (flattened vertebral bodies).
- French-telephone femurs.
- H-shaped vertebrae.
- Cloverleaf skull (severe craniosynostosis).

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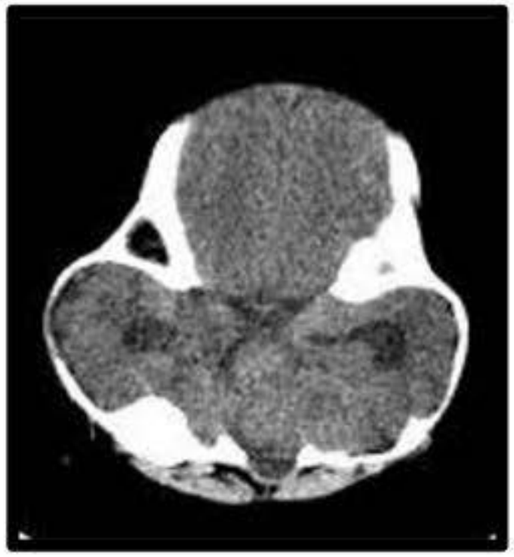
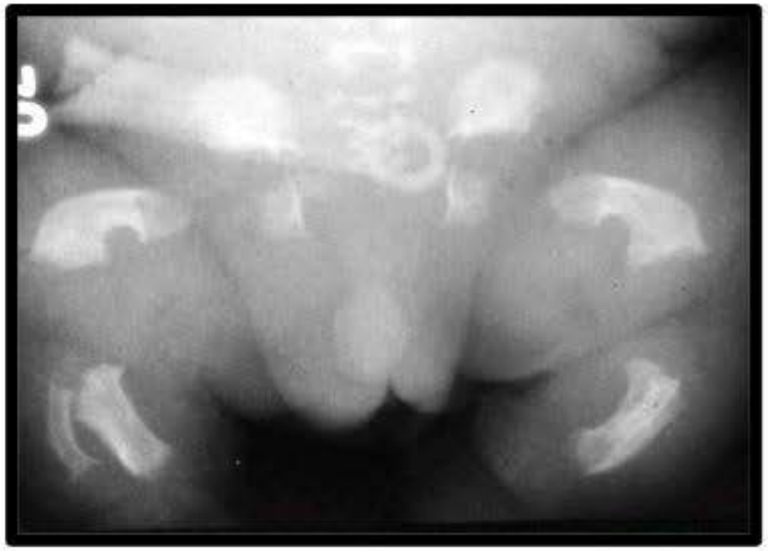
❖ **Mode of inheritance?**
- ✓ Autosomal Dominant
- ✓ Defect in FGFR-3

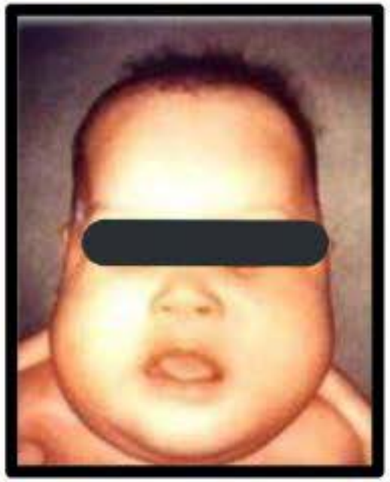
❖ **Most common cause of death?**
- ✓ Craniocervical junction compression
- ✓ Pulmonary hypoplasia

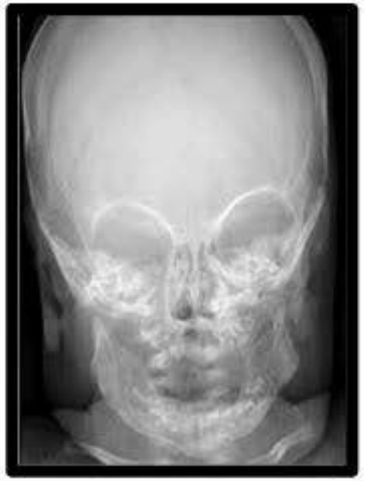


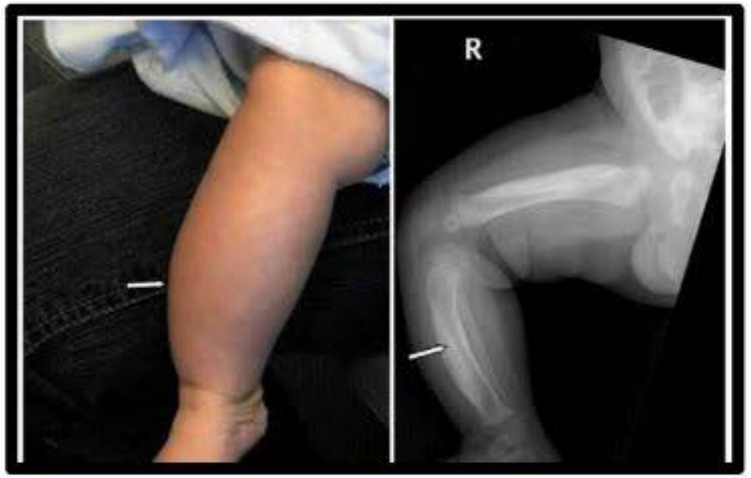
❖ **Types**
- Type I
  - Without cloverleaf skull
  - Telephone receiver femur
- Type II
  - With cloverleaf skull
  - Straight femur



22) Infantile cortical hyperostosis (Caffey Disease)







The bone changes typically begin before six months of age and resolve by two years.

Clinical features

- Extreme irritability.
- Soft tissue swelling.
- Fever.
- Anorexia.
- Cortical thickening of bones.
- Pseudoparalysis

Differential diagnosis

- Osteomyelitis
- Hypervitaminosis A.
- Child abuse.
- Leukemia

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❖ **Most common affected bone?**

✓ **Mandible**

❖ **Mode of inheritance?**

✓ Autosomal dominant

✓ COL1A1 gene

❖ **Laboratory finding ?**

✓ Leukocytosis

✓ Thrombocytosis

✓ High ESR

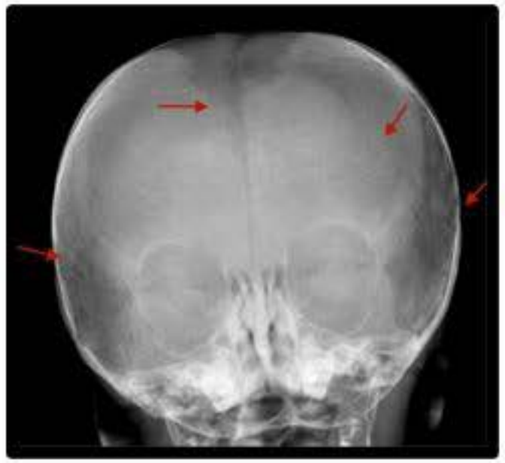
✓ High Alkaline phosphatase

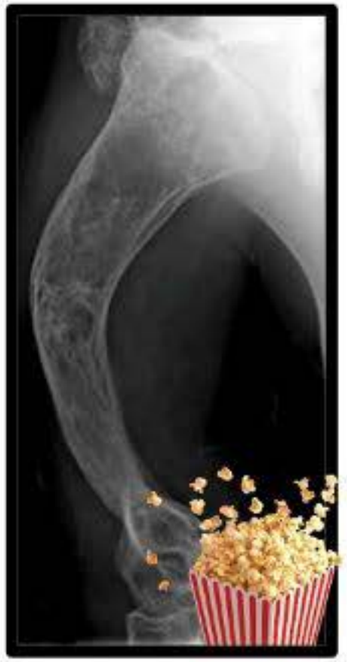
❖ **Prognosis**

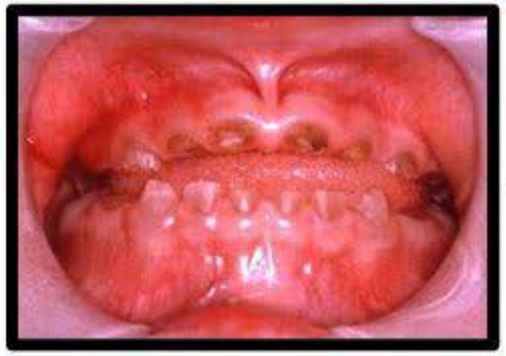
✓ Self-limited condition

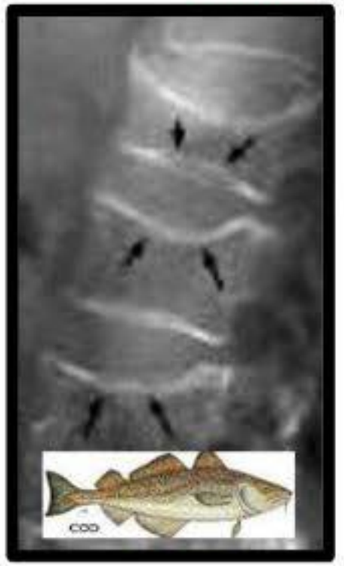
✓ Deformities of the involved bones.

23) Osteogenesis Imperfecta









Clinical features

- Short stature
- Scoliosis
- Blue sclera
- Dentinogenesis imperfecta
- Hyperextensible joints
- Easy bruising.
- Wormian bones (small, irregular bones along the cranial sutures)
- Codfish vertebrae
- Flared metaphysis (popcorn-like)

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❖ Mode of inheritance?

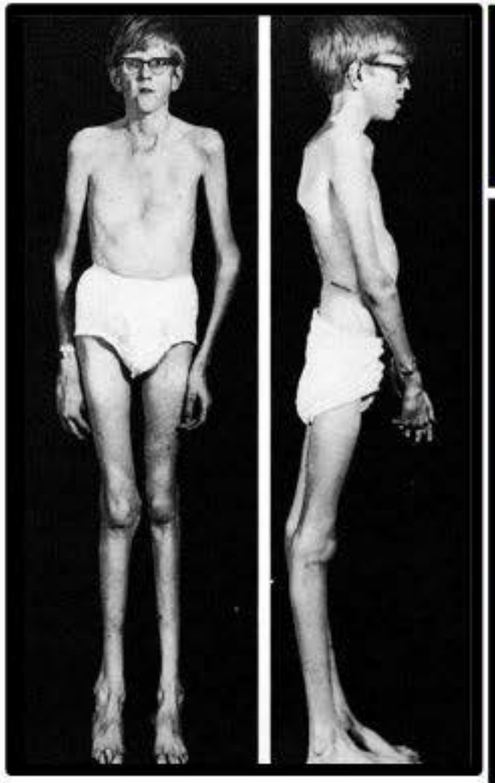
- ✓ AD (I – IV)
- ✓ AR (rare types + IIB)
- ✓ COL1A1 gene

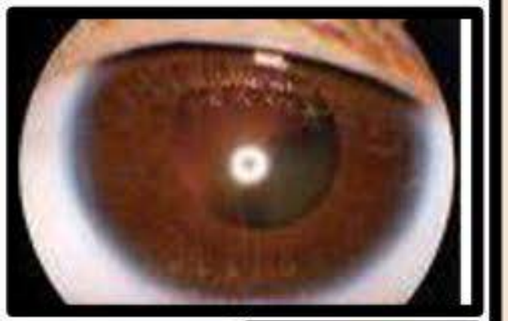
❖ Common Types

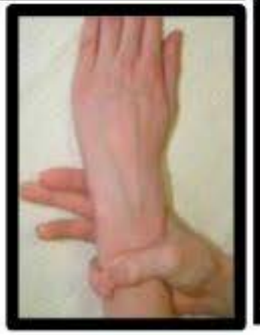
1. Type I
   - Most common
   - Mildest form
   - Classic nondeforming
   - Hearing loss in 50%
2. Type II
   - Most severe
   - Death during newborn
3. Type III
   - Progressive deforming
   - Hydrocephalus
4. Type IV
   - Normal sclera
   - Tibial bowing (hallmark)

❖ Laboratory finding ?

- ✓ High alkaline phosphatase
- ✓ Hypercalciuria







24) Marfan Syndrome

Clinical features

Major criteria:

- ✓ Ectopia lentis.
- ✓ Aortic dilatation or dissection.
- ✓ Family history

Systematic score features :

Wrist AND thumb sign	3
Wrist OR thumb sign	1
Pectus carinatum deformity	2
Pectus excavatum or chest asymmetry	1
Hindfoot deformity	2
Pes planus	1
Pneumothorax	2
Dural ectasia	2
Protrusio acetabuli	2
Reduced upper segment/lower segment ratio AND increased arm/height ratio AND no severe scoliosis	1
Scoliosis or thoracolumbar kyphosis	1
Reduced elbow extension	1
Facial features (3/5): dolichocephaly, enophthalmos, downslanting palpebral fissures, malar hypoplasia, retrognathia	1
Skin striae	1
Myopia >3 diopters	1
Mitral valve prolapse	1

❖ Mode of inheritance?

- ✓ Autosomal dominant
- ✓ FBN1 gene

❖ Monitoring?

- ✓ Annual echocardiogram.

How to confirm the diagnosis?

Ghent criteria

- 2 major criteria
- 1 major + FBN1 gene mutation
- 1 major + ≥7 more systematic score

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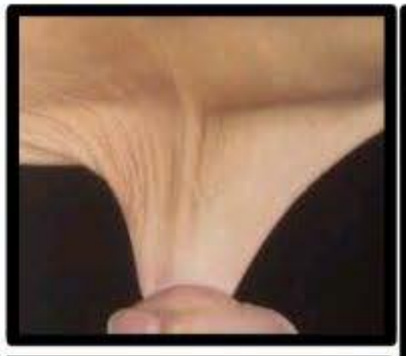
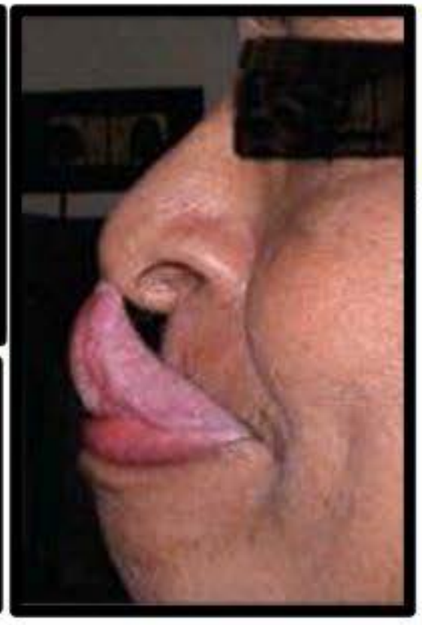
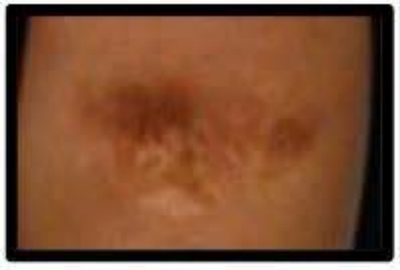
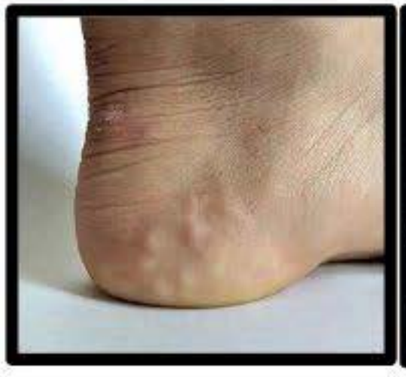
❖ Most common cardiac defect ?

- ✓ **Mitral valve prolapse**
- ✓ Aortic root dilatation

❖ Most common cause of death?

- ✓ Cardiovascular complications
- ✓ Rupture aortic root dilatation


25) Ehlers–Danlos syndromes (EDS)

Clinical features

- ✓ Generalized joint hypermobility
- ✓ Hyperextensible skin
- ✓ Fragile skin.
- ✓ Increased bruising
- ✓ Velvety skin
- ✓ Kyphoscoliosis
- ✓ Swan-neck malformation
- ✓ Piezogenic papules
- ✓ Gorlin's sign
- ✓ Cigarette paper scar
- ✓ Fish mouth appearance of skin tears.
- ✓ Hypotonia

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THE BEIGHTON SCORE

How to Assess Joint Hypermobility

A numerical mobility score of 0 to 9, one point allocated for the ability to perform each of the following tests:

Pull little finger back beyond 90°
(one point for each side)

Pull thumb back to touch forearm
(one point for each side)

Bend elbow backwards beyond 10°
(one point for each side)

Bend knee backwards beyond 10°
(one point for each side)

Lie hands flat on floor while keeping knees straight and bending forward at waist

A positive Beighton score for adults is 5 out of the 9 possible points; for children, a positive score is at least 6 out of 9 points.

- ❖ **Mode of inheritance?**
- ✓ Autosomal dominant
- ✓ Rarely AR

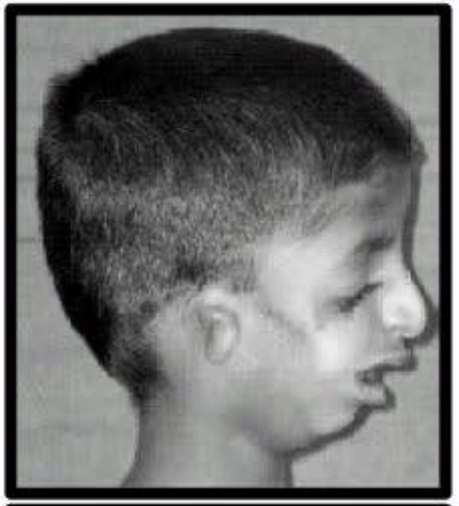
- ❖ **Most common cardiac defect ?**
- ✓ Mitral valve prolapse
- ✓ Aortic root dilatation

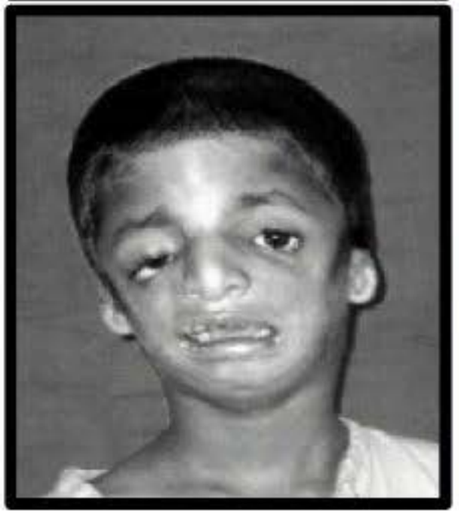
- ❖ **Laboratory finding include normal coagulation expect ?**
- ✓ Capillary fragility test

- ❖ **Monitoring?**
- ✓ Annual echocardiogram.

- ❖ **Most severe type?**
- ✓ Vascular (vEDS; EDS type IV)

26) Treacher Collins Syndrome (Mandibulofacial Dysostosis)





Clinical features

- ✓ Zygomatic and mandibular bone hypoplasia
- ✓ Eyelid colobomas
- ✓ Sparse eye lashes
- ✓ Microretrognathia
- ✓ Choanal atresia
- ✓ Microtia
- ✓ Conductive hearing loss
- ✓ Cleft lip / palate

Pediatric On Squores
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❖ **Most common neonatal problem?**

- ✓ Airway difficulties

❖ **What is the level of intelligence ?**

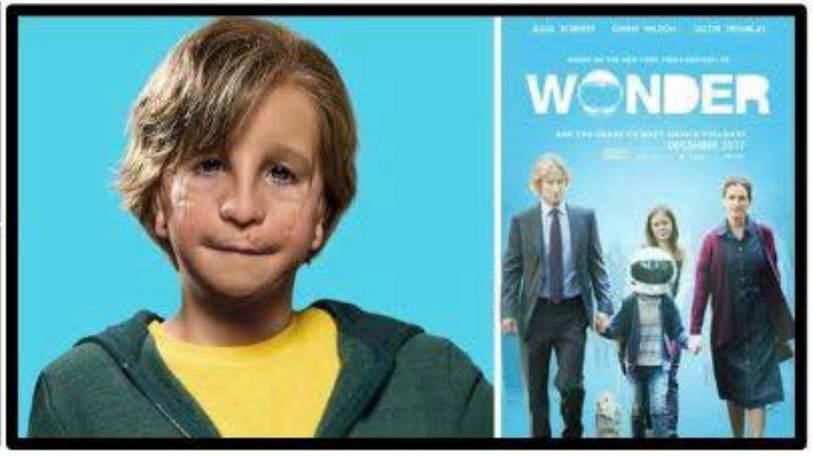
- ✓ Normal

❖ **Mode of inheritance?**

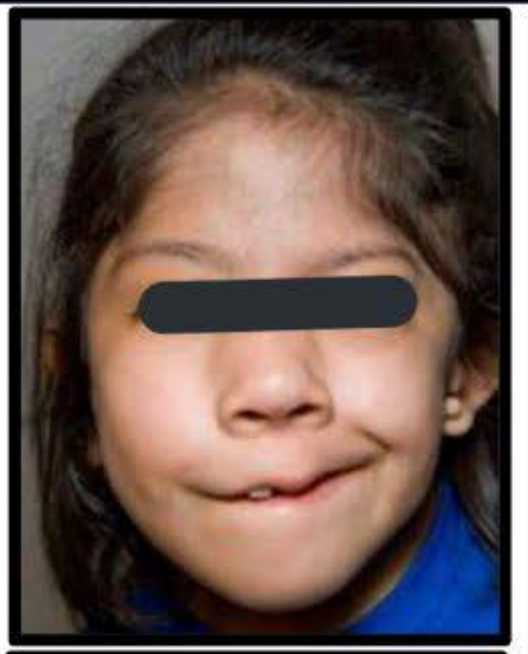
- ✓ Autosomal dominant
- ✓ TCOF1 gene

❖ **Clinic follow up ?**

- ✓ Vision assessment
- ✓ Hearing assessment
- ✓ Feeding difficulties
- ✓ Speech therapy
- ✓ Psychosocial therapy



27) Goldenhar syndrome (oculo-auriculo-vertebral spectrum)



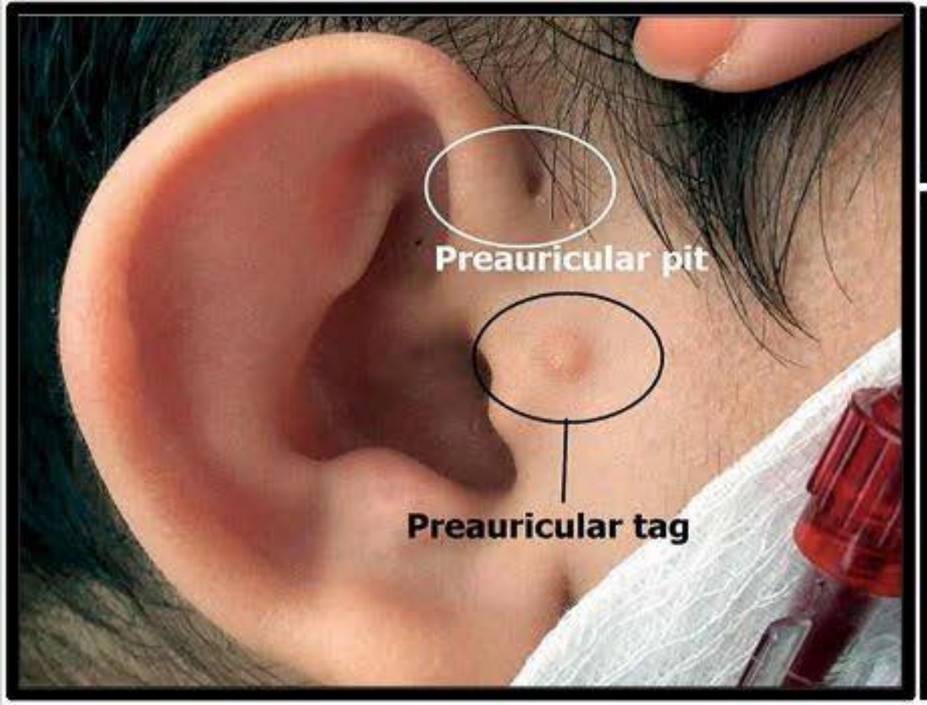
Clinical features

- ✓ Facial asymmetry (hemifacial macrosomia)
- ✓ Microtia
- ✓ Preauricular tags
- ✓ Microphthalmia
- ✓ Limbal (Epibulbar) dermoid (Pathagnomic)
- ✓ Eyelid coloboma
- ✓ Facial nerve involvement leads to hypoplasia of the facial muscles.
- ✓ Hypoplasia or absence of the parotid gland.
- ✓ Vetebral anomalies (Hemivertebrae)
- ✓ Cardiac defect (VSD)

❖ **Mode of inheritance?**

- ✓ Sporadic
- ✓ Abnormal 1st and 2nd brachial arches

28) Branchio-oto-renal (BOR) syndrome



Clinical features

- ✓ Branchial cleft fistulas / cysts
- ✓ Preauricular pits / tags
- ✓ Renal aplasia / hypoplasia
- ✓ Sensory / conductive hearing loss
- ✓ Pulmonary hypoplasia

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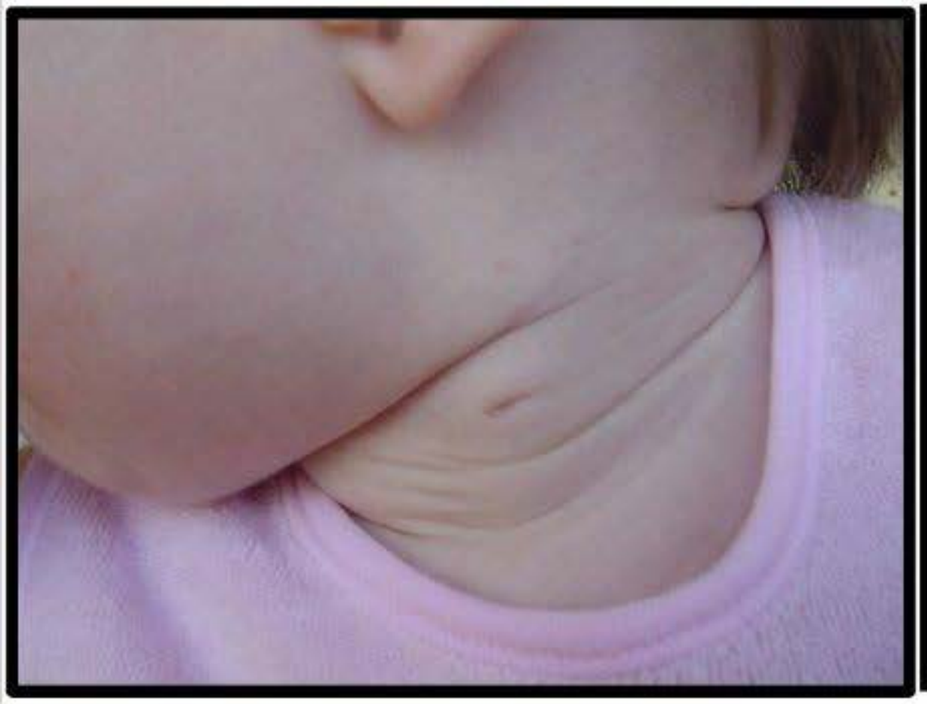
❖ **Mode of inheritance?**

✓ Autosomal dominant

❖ **Laboratory finding ?**

- ✓ Low renin
- ✓ Low aldosterone
- ✓ Hyperkalemia
- ✓ Hyperchloremia
- ✓ Metabolic acidosis



Major criteria
Deafness
Preauricular pits
Renal anomalies
Minor criteria
External ear anomalies
Middle ear anomalies
Inner ear anomalies
Preauricular tags
Others: Facial asymmetry, palatal abnormalities

Three major or two major and two minor criteria are required for a diagnosis of Branchio-oto-renal syndrome. In those with an affected first-degree relative with BOR syndrome, one major criterion is sufficient for diagnosis.

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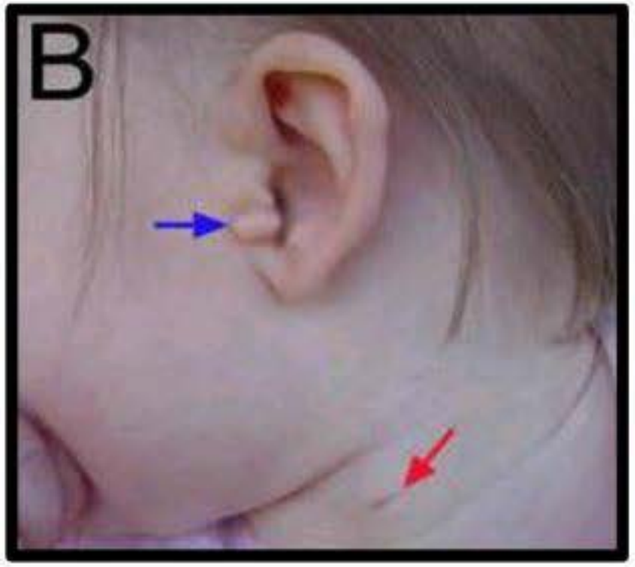
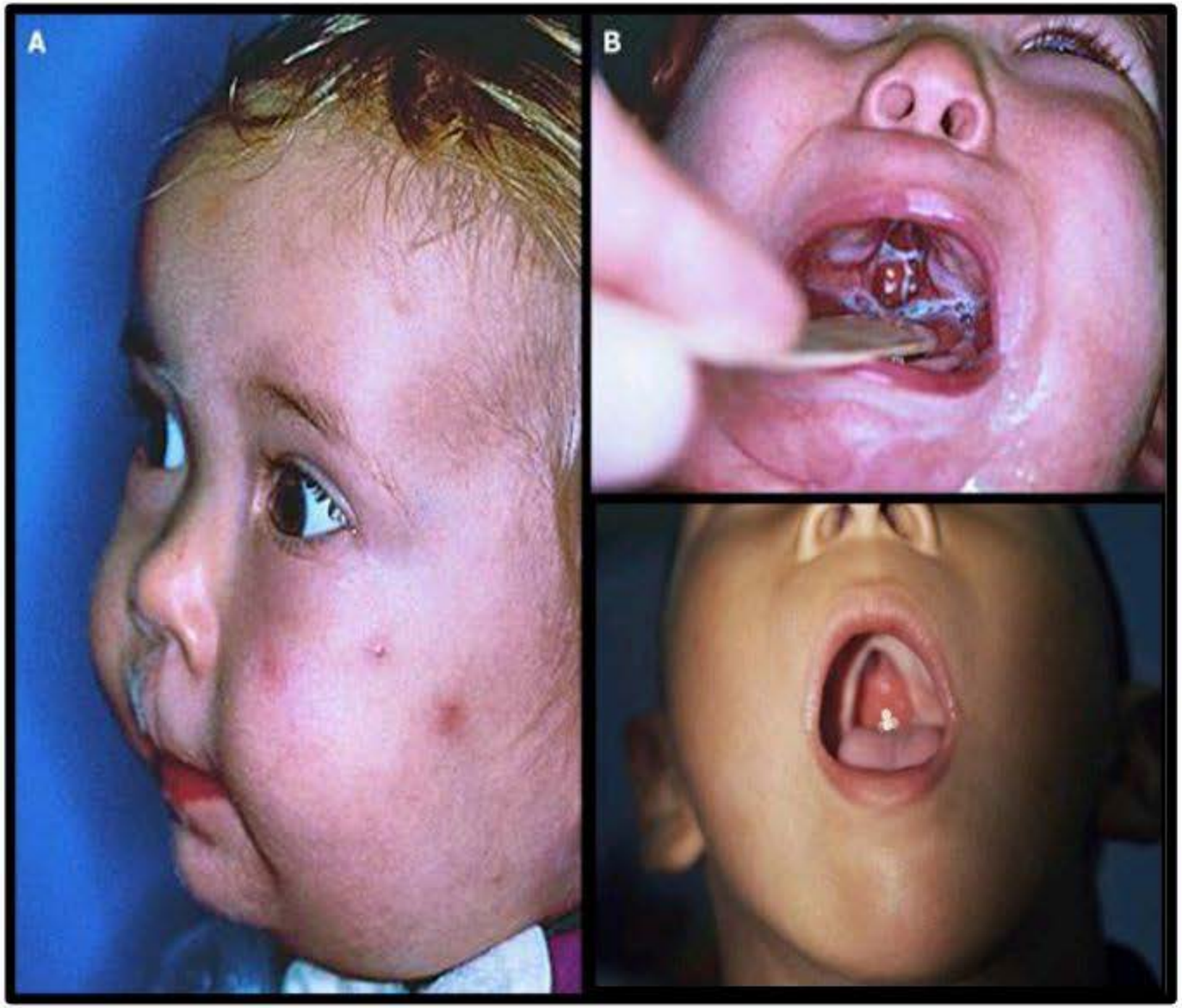


29) Pierre Robin sequence (Pierre Robin syndrome)



Clinical features

- ✓ Mandibular hypoplasia
- ✓ Micrognathia
- ✓ Glossoptosis (backward displacement of the tongue)
- ✓ U shaped cleft palate
- ✓ Birdlike facial appearance.
- ✓ Upper airway obstruction

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❖ **Mode of inheritance?**

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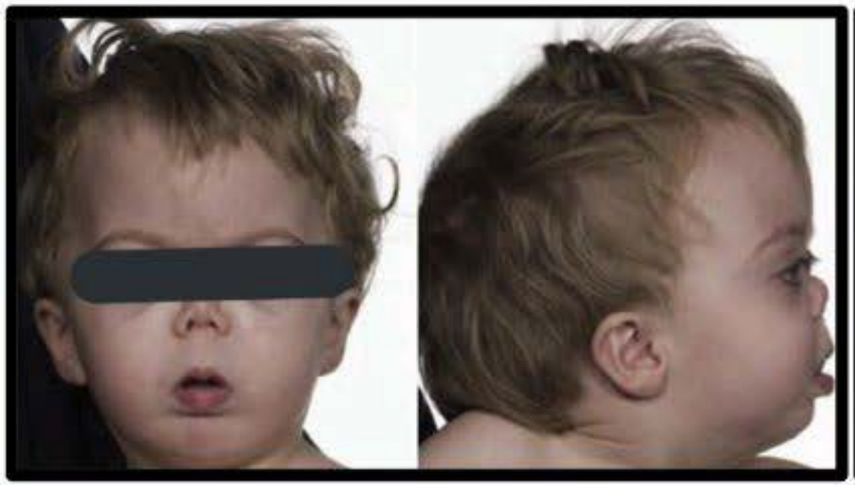
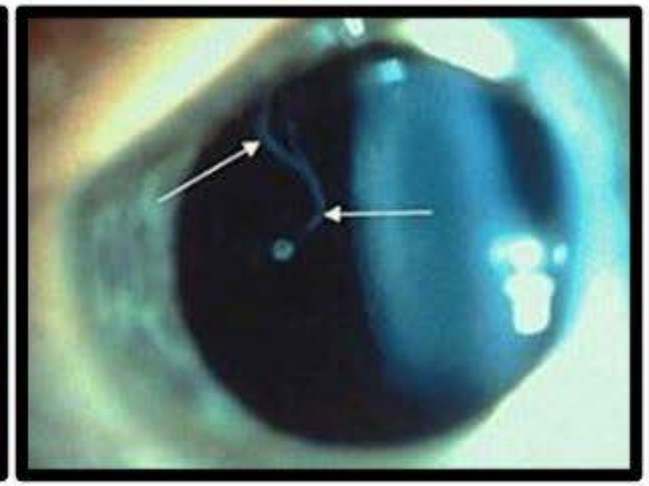
- ✓ De novo mutation
- ✓ SOX9 gene

❖ **Most common cause of death?**

- ✓ Upper airway obstruction

30) Stickler syndrome (hereditary arthroophthalmopathy)




Clinical features

- ✓ Pierre Robin sequence +
- ✓ Flat midface and nasal bridge
- ✓ Joint hypermobility
- ✓ Myopia
- ✓ Sensorineural deafness
- ✓ Cleft of the soft palate
- ✓ Arthritis

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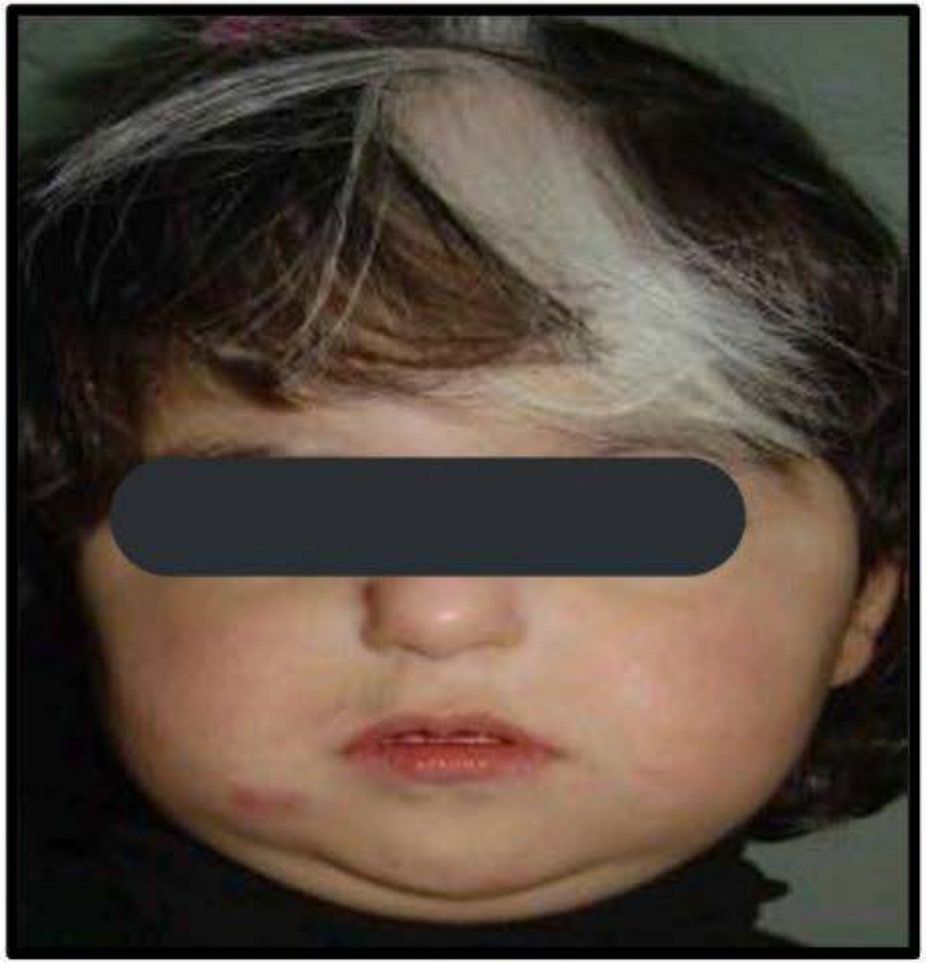
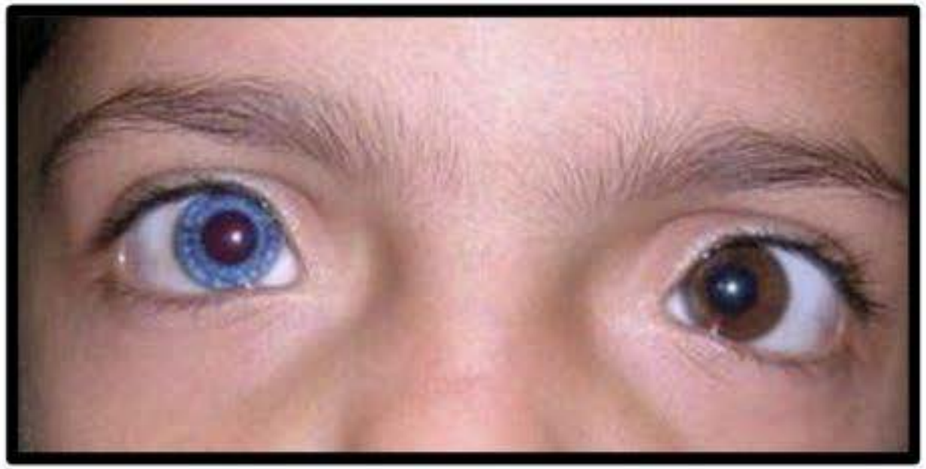
❖ **Mode of inheritance?**
- ✓ Autosomal dominant

❖ **What is the pathognomonic eye finding?**
- ✓ Vitreous gel anomaly

❖ **What is ophthalmic emergency associated with stickler syndrome?**
- ✓ Retinal detachment

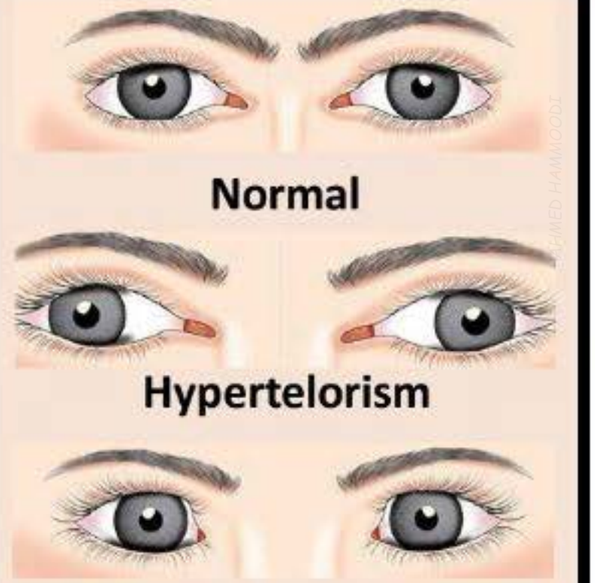




31) Waardenburg syndrome

Clinical features

- ✓ White forelock
- ✓ Square face
- ✓ Thin nose, wide nasal bridge
- ✓ Dystopia canthorum
- ✓ Heterochromia
- ✓ Sensorineural deafness
- ✓ Skin depigmentation



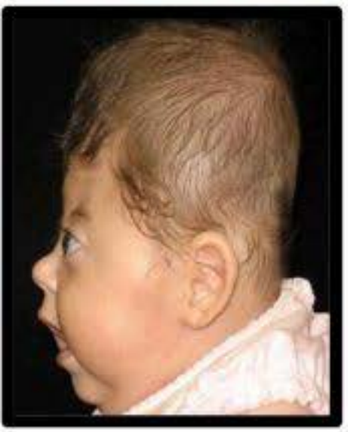
❖ **Mode of inheritance?**

- ✓ Autosomal dominant

❖ **Most common gastrointestinal association?**

- ✓ Hirschsprung disease

32) Apert syndrome



❖ **Clinical features**

- ✓ Complex syndactyly (mitten hand)
- ✓ Bicoronal craniosynostosis
- ✓ Maxillary hypoplasia
- ✓ Exorbitism (Protruding eyes)
- ✓ Hypertelorism
- ✓ Midface hypoplasia
- ✓ Intellectual disability

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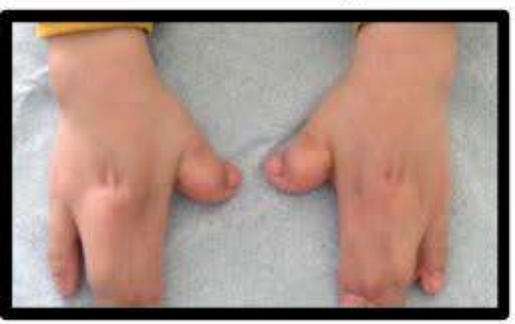
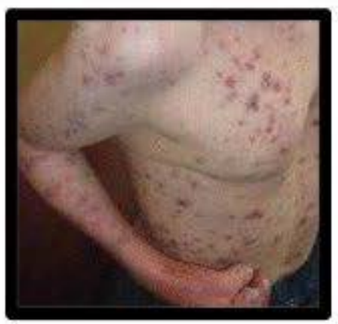
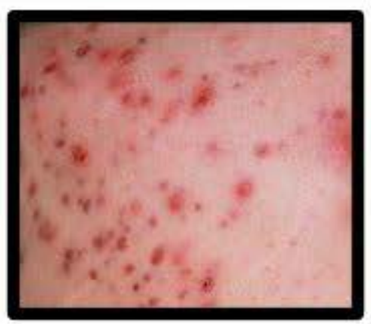
❖ **What is the most common dermatological complication with Apert syndrome ?**

- ✓ Severe acne vulgaris







Other Craniosynostosis syndromes

❖ **Craniosynostosis syndromes**

1. Apert syndrome (Bicoronal suture)
2. Crouzon syndrome (Bicoronal suture)
3. Carpenter syndrome (Multiple sutures)
4. Saethre-Chotzen syndrome (Multiple sutures)
5. Pfeiffer syndrome (Multiple sutures)

❖ **Mode of inheritance?**

- ✓ All of them Autosomal dominant except :
- ✓ Carpenter syndrome (Autosomal recessive)

❖ **Which craniosynostosis syndromes have normal intelligence?**

- ✓ Crouzon syndrome

❖ **Which craniosynostosis syndromes have normal hands?**

- ✓ Crouzon syndrome (No syndactyly)

❖ **Which craniosynostosis syndromes without hypertelorism?**

- ✓ Saethre-Chotzen syndrome

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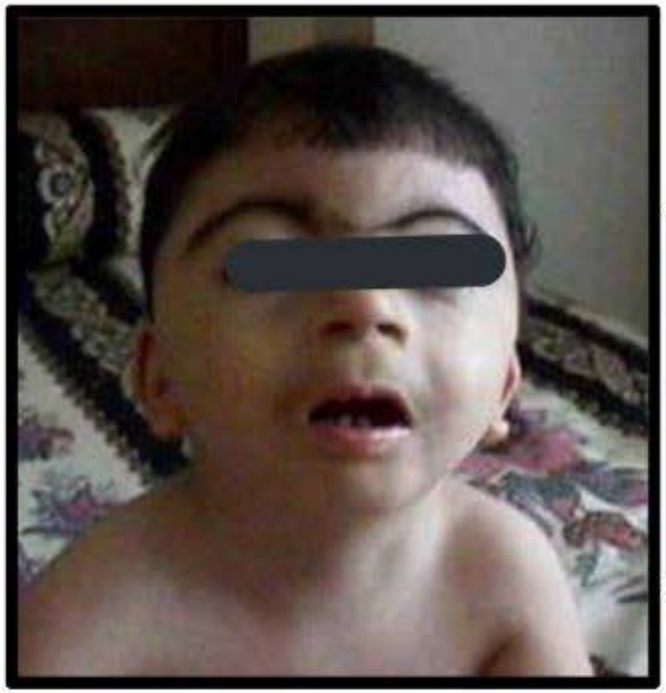
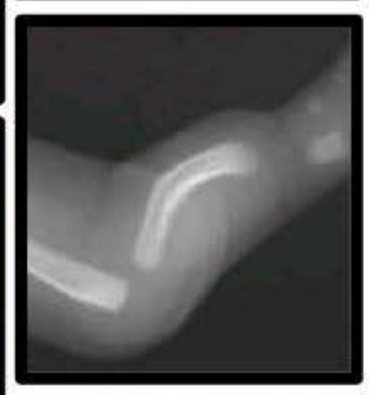
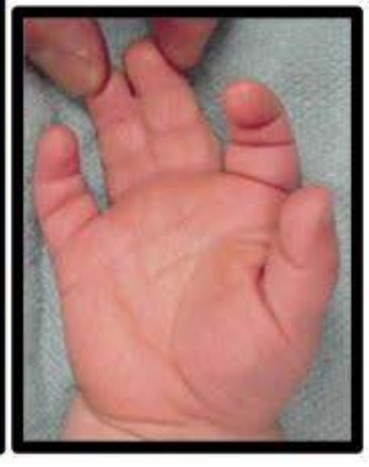
33) Cornelia de Lange Syndrome







❖ Clinical features

- ✓ Long eyelashes
- ✓ Bushy and arched eyebrows
- ✓ Hirsutism
- ✓ Low hairline
- ✓ Downward-turned mouth
- ✓ IUGR
- ✓ Short stature
- ✓ Thin upper lip
- ✓ Micromelia
- ✓ Lobster hand
- ✓ Syndactyly
- ✓ Ulnar dysplasia
- ✓ Mental retardation

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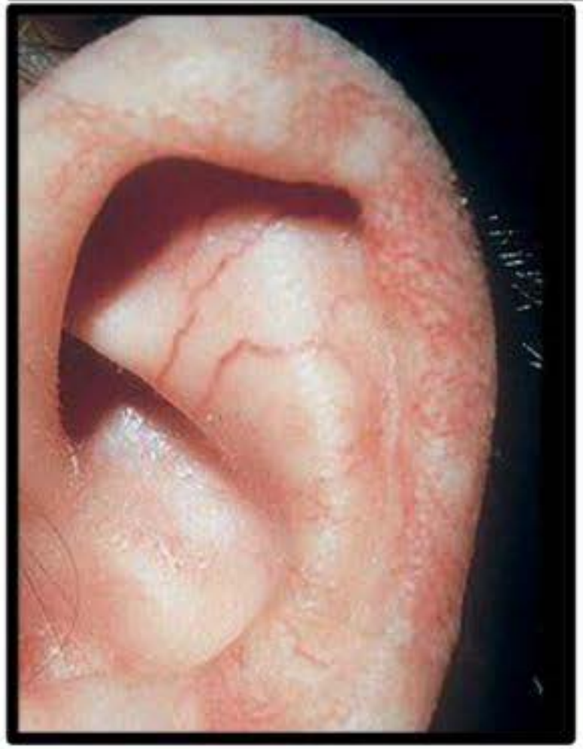
❖ **Mode of inheritance?**

- ✓ Autosomal dominant

❖ **Most common cardiac defect ?**

- ✓ Pulmonary stenosis
- ✓ VSD
- ✓ ASD
- ✓ CoA

34) Ataxia Telangiectasia



❖ Clinical features

- ✓ Cerebellar ataxia
- ✓ Oculocutaneous telangiectasia
  - Bulbar conjunctivae.
  - Ears
  - Neck
  - Cubital fossae
- ✓ Recurrent infection
- ✓ Increase risk of malignancy

❖ **Laboratory finding ?**

- ✓ High serum alpha-fetoprotein (AFP)
- ✓ High carcinoembryonic antigen (CEA)
- ✓ Low IgA, IgG & IgE

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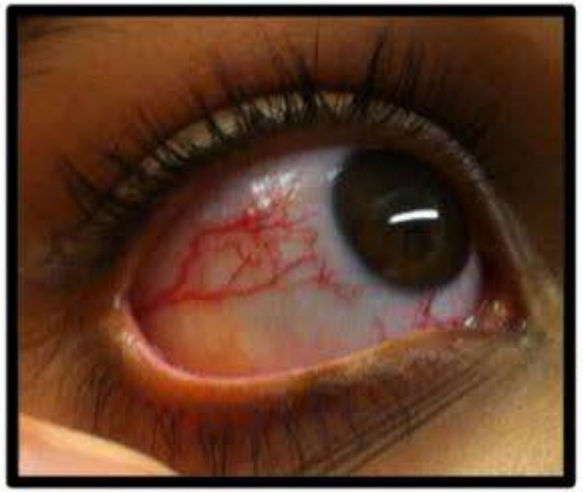
❖ **What is the most consistent laboratory abnormality?**

- ✓ High AFP



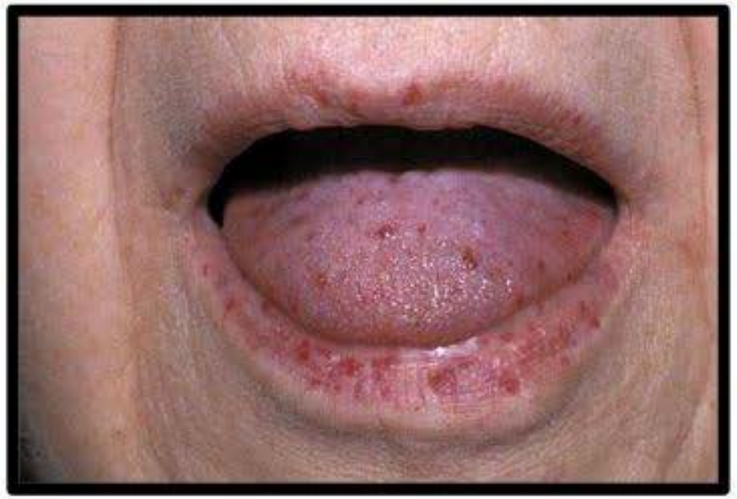
❖ **Mode of inheritance?**

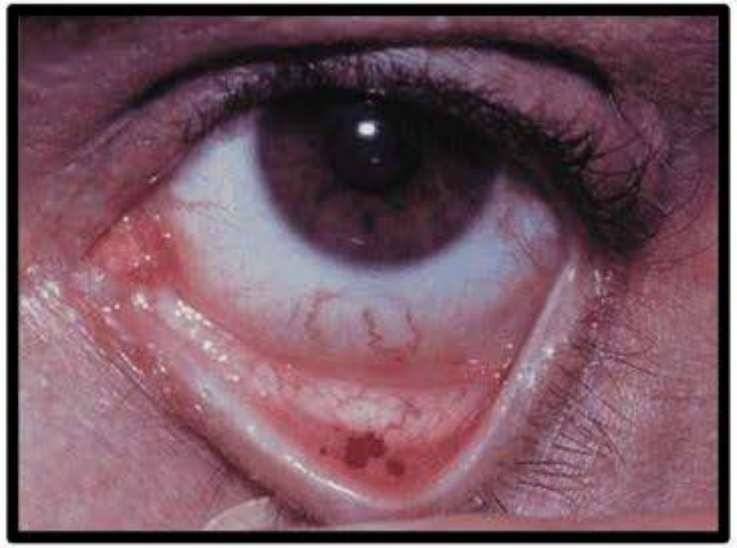
- ✓ Autosomal recessive
- ✓ ATM gene
- ✓ Due to chromosome instability

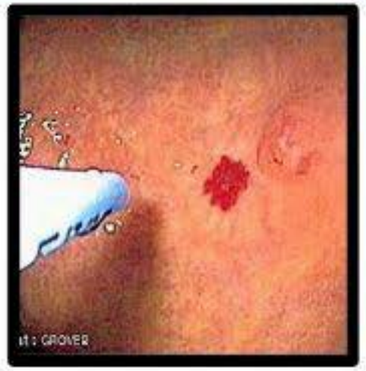
❖ **Which part of immune system is impaired?**

- ✓ Both cellular and humoral immunity

- Associated with increase sensitivity to ionizing radiation







35) Osler Weber Rendu Syndrome (Hereditary hemorrhagic Telangiectasia)

❖ Clinical features

- ✓ Telangiectasia (lips, gingiva, tongue, palate and palpebral conjunctiva)
- ✓ Recurrent epistaxis (Night time)
- ✓ Vascular malformation (Lung, liver, GI & brain)

❖ Mode of inheritance?

- ✓ Autosomal dominant

❖ Complications ?

- ✓ Intracranial hemorrhage
- ✓ Pulmonary hemorrhage
- ✓ GI bleeding

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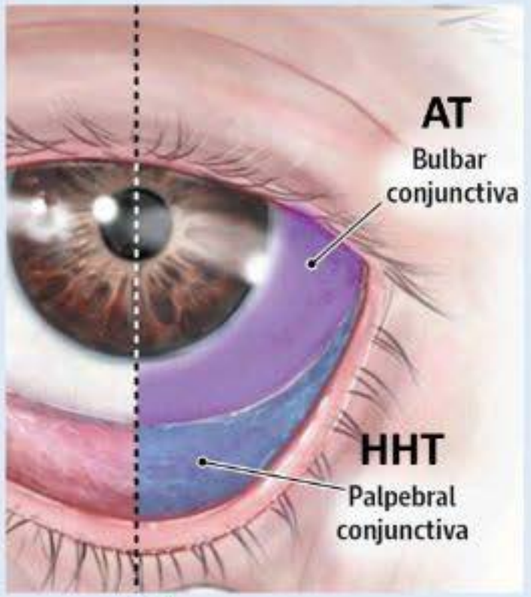
The most common cause of pulmonary arteriovenous malformation

Conjunctival finding in Hereditary hemorrhagic Telangiectasia (HHT)

VS

Ataxia Telangiectasia (AT)



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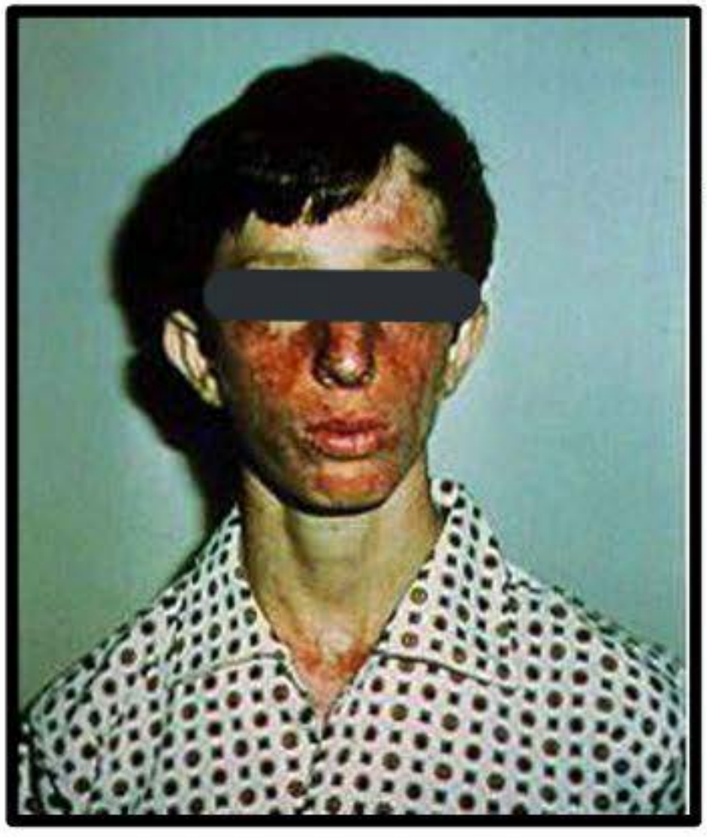

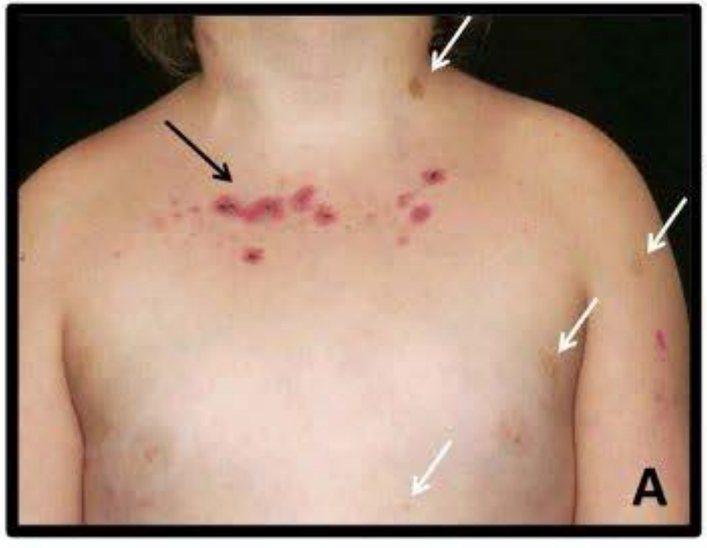

36) Bloom syndrome

❖ Clinical features

- ✓ Café-au-lait spots
- ✓ Photosensitive rash
- ✓ Long narrow face
- ✓ Prominent nose
- ✓ IUGR
- ✓ Microcephaly
- ✓ Short stature
- ✓ Infertility
- ✓ Immune deficiency

❖ Mode of inheritance?

- ✓ Autosomal recessive
- ✓ Due to chromosome instability

❖ Increase risk of:

- ✓ Malignancies
- ✓ Immunodeficiency

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❖ Laboratory finding ?

- ✓ Low immunoglobulin level
- ✓ Lymphopenia

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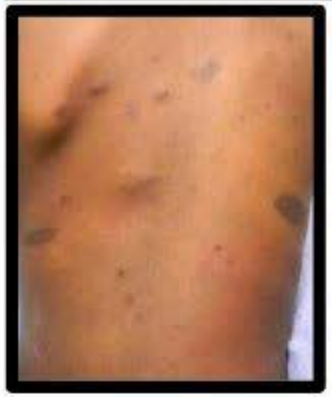
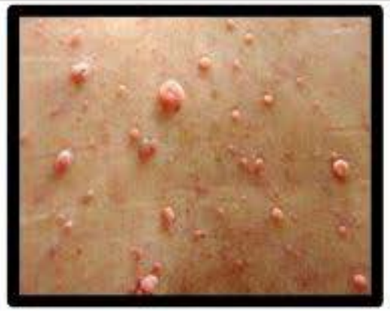
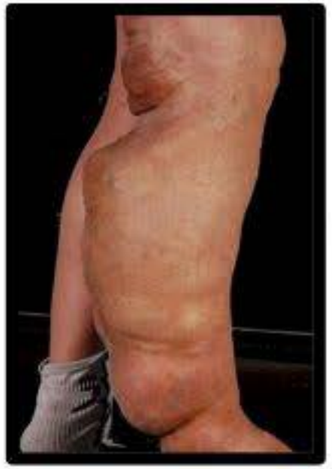
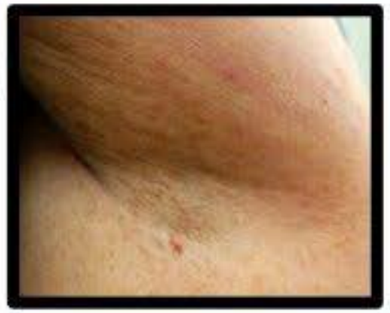
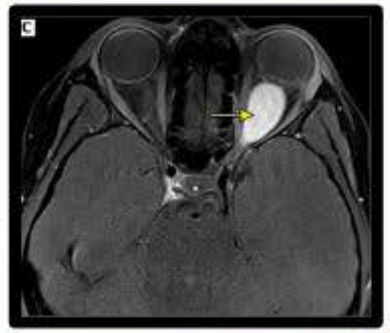
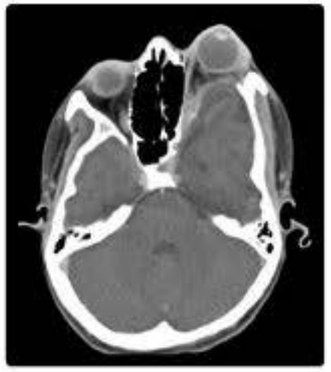
❖ Most common cause of death?

- ✓ Cancers
  - Mainly leukemia

Avoid radiographs
Adequate sun protection

37) Neurofibromatosis type 1 (von Recklinghausen disease)


❖ Diagnostic criteria (≥ 2 of the following)

- ✓ ≥ 6 café-au-lait macules
  - ▪ > 5 mm in in prepubertal
  - ▪ > 15 mm in in postpubertal
- ✓ ≥ 2 neurofibromas or 1 plexiform neurofibroma
- ✓ Axillary or inguinal freckling (Crowe sign)
- ✓ Optic glioma
- ✓ ≥ 2 iris hamartoma (Lisch nodules)
- ✓ Distinctive bony lesion:
  - ▪ Sphenoid dysplasia, or
  - ▪ Tibial pseudoarthrosis, or
  - ▪ Cortical thickening of the long bone
- ✓ A first-degree relative with NF1

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❖ What is 1st manifestation typically appears ?

- ✓ Café-au-lait macules

❖ What is most common type of benign tumor in NF1 ?

- ✓ Neurofibromas

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❖ Mode of inheritance?

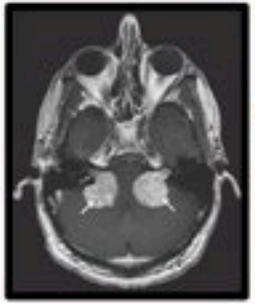
- ✓ AD (50%)
- ✓ De novo (50%)
- ✓ NF1 gene on chromosome 17

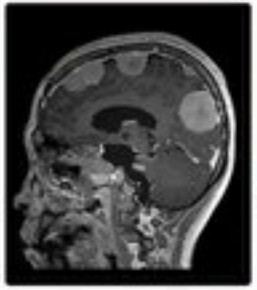
❖ Increases risk of :

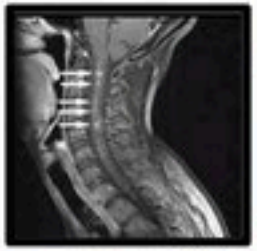
- ✓ **Malignancies**
- ✓ Primary HTN
- ✓ Learning disabilities
- ✓ Seizures

❖ Follow up :

- ✓ Annual vision exam
- ✓ BP monitoring
- ✓ Annual full physical examination
- ✓ Tumor surveillance







38) Neurofibromatosis type 2 (MISME syndrome)

❖ **Diagnostic criteria** (1 of the following sets)

- ✓ Bilateral vestibular schwannomas (acoustic neuromas)
- ✓ Unilateral vestibular schwannoma AND first-degree relative with NF2.
- ✓ Unilateral vestibular schwannoma Or first-degree relative with NF2 AND any 2 of the following:
  - Meningioma
  - Nonvestibular schwannoma
  - Ependymoma
  - Cataract

Other clinical features

- ✓ Hearing loss
- ✓ Tinnitus

❖ **What is 1st manifestation typically appears ?**

✓ Cataract

❖ **What is most common type of tumor in NF2?**

✓ Schwannomas

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❖ **What is most common affected cranial nerve with schwannomas?**

✓ CN VIII (vestibulocochlear nerve)

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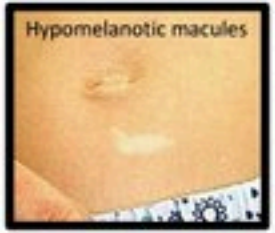
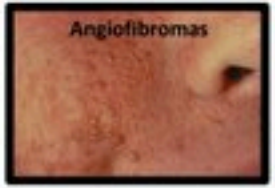
❖ **Mode of inheritance?**

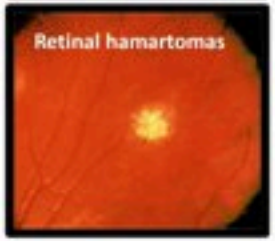
- ✓ Autosomal dominant
- ✓ NF2 gene on chromosome 22

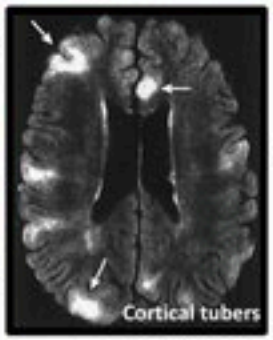
- Café-au-lait macules can be seen but are much less frequent in NF2.
- NF2 is not associated with the cognitive impairment that is often seen with NF1.

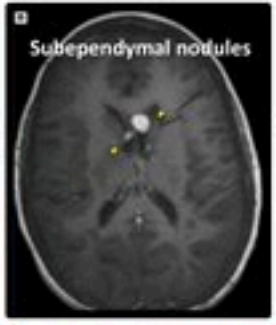
39) Tuberous sclerosis complex (TSC)

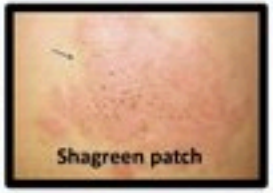


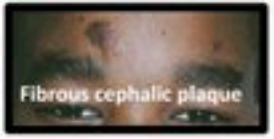


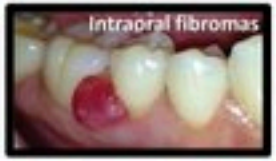


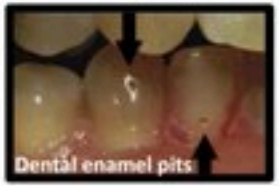


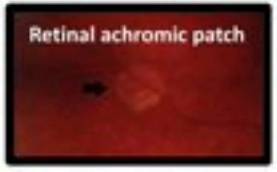












❖ Diagnostic criteria

Major clinical features:

1. ≥ 3 Hypomelanotic (Ash leaf) spot >5 mm
2. ≥3 Angiofibromas or fibrous cephalic plaque
3. ≥2 Ungual fibromas (≥2)
4. Shagreen patch (connective tissue nevus)
5. Multiple retinal hamartomas
6. Cortical dysplasias (cortical tubers)
7. Subependymal nodules
8. Subependymal giant cell astrocytoma
9. Cardiac rhabdomyoma
10. Lymphangioleiomyomatosis (LAM)*
11. Angiomyolipomas (≥2)*

Minor clinical features:

12. "Confetti" skin lesions
13. Dental enamel pits (≥3)
14. Intraoral fibromas (≥2)
15. Retinal achromic patch
16. Multiple renal cysts
17. Nonrenal hamartomas

Definite TSC: 2 major or 1 major and ≥ 2 or
Possible TSC: 1 major or ≥ 2 minor

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❖ **What is the most common cardiac defect?**
✓ Rhabdomyoma

❖ **What is the most common renal manifestation ?**
✓ Angiomyolipomas

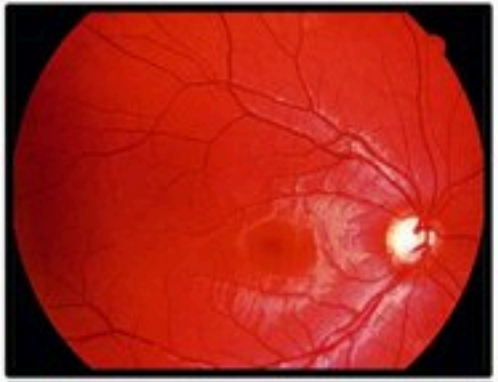
❖ **Mode of inheritance?**
✓ Autosomal dominant
✓ TSC1 gene on ch. 9
✓ TSC2 gene on ch. 16

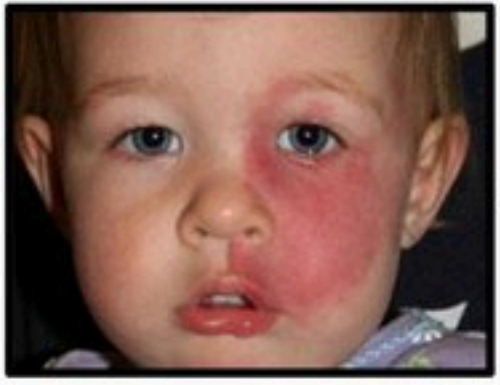
❖ **Which 2 major criteria without other features does not meet criteria for definite diagnosis?**
✓ Angiomyolipomas
✓ LAM

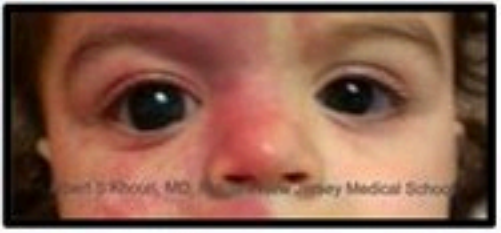
❖ **What is the most common neurological complication ?**
✓ Infantile spasm
➢ 1st Tx Vigabatrin

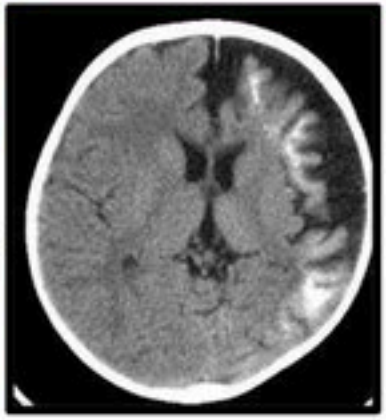
❖ **TSC FDA approved Tx?**
✓ Everolimus

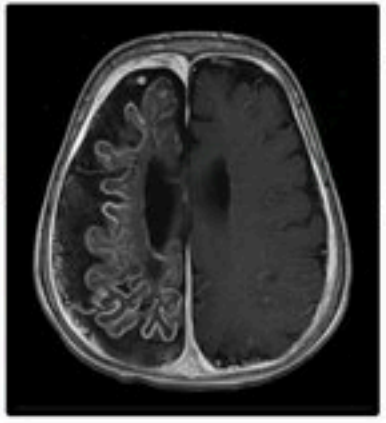
40) Sturge Weber syndrome











❖ Clinical features

- ✓ Port wine stain (1st & 2nd division of trigeminal nerve)
- ✓ Ipsilateral leptomeningeal angioma
- ✓ Glaucoma
- ✓ Diffuse choroidal hemangioma
- ✓ Seizures (on contralateral side)
- ✓ Focal neurologic deficits
- ✓ Buphthalmos
- ✓ Intellectual disability.

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❖ Mode of inheritance?

- ✓ Not inherited, due somatic mutation

❖ What is the best neuroimaging diagnostic study?

- ✓ Brain MRI with gadolinium contrast
- ✓ Tram track calcification

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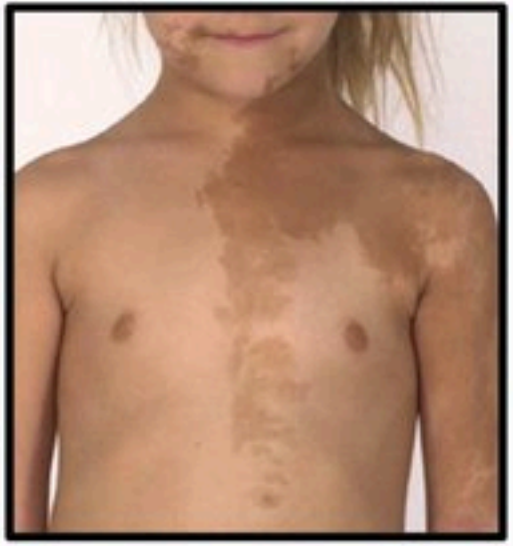
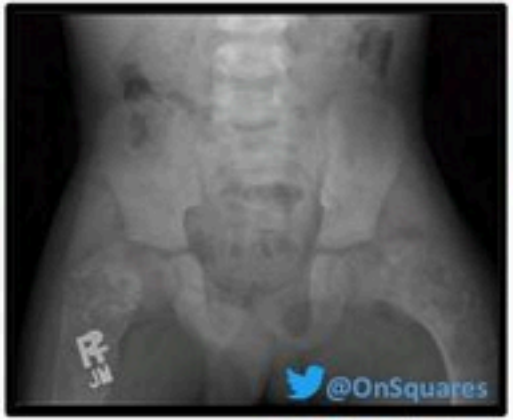




41) McCune-Albright syndrome (MAS)

❖ Clinical features

Classic triad

- ✓ Precocious puberty
- ✓ Café-au-lait macules
- ✓ Polyostotic fibrous dysplasia

❖ What is the most characteristic laboratory finding?

- ✓ High growth hormone
- ✓ High prolactin
- ✓ High luteinizing hormone (LH)
- ✓ High follicle-stimulating hormone (FSH)
- ✓ Hypophosphatemia & hyperphosphaturia

❖ What is the classic presenting sign of Precocious puberty in MAS?

- ✓ Vaginal bleeding

❖ How to differentiated café-au-lait macules from those of neurofibromatosis (NF)

In MAS

- ✓ Coast of Maine borders
- ✓ Irregular borders
- ✓ Never cross midline

In NF

- ✓ Coast of California
- ✓ Smooth borders
- ✓ Cross midline

❖ What is the other common endocrine association in MAS?

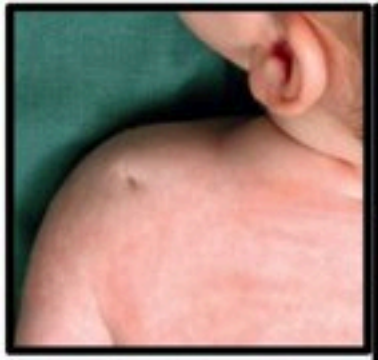
- ✓ Hyperthyroidism

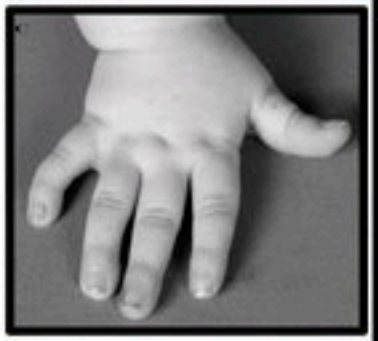
❖ Mode of inheritance?

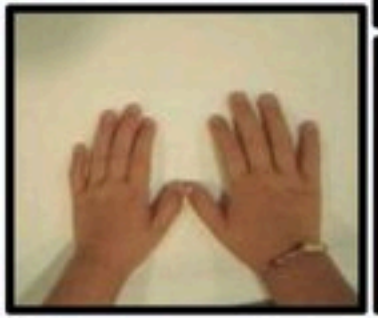
- ✓ Sporadic
- ✓ Mutation in the GNAS1 gene
- ✓ More common in girls

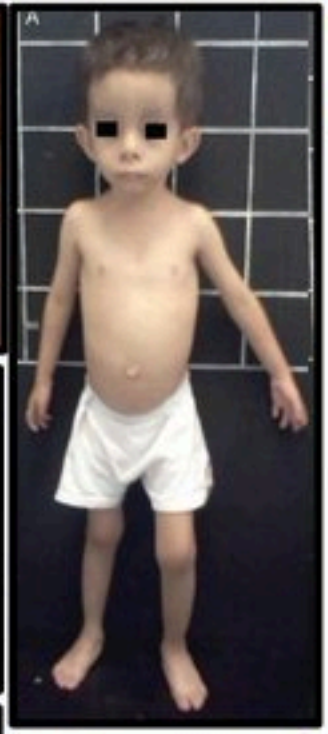
❖ Treatment

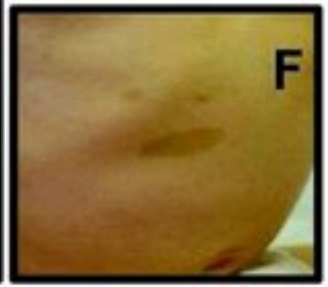
- ✓ GnRH agonist
- ✓ Oral phosphate and calcitriol











42) Russel Silver syndrome

❖ Clinical features

- ✓ Short stature
- ✓ Small for gestational age
- ✓ Hemihypertrophy
- ✓ Pseudohydrocephlus
- ✓ Triangular face
- ✓ Inverted V shaped mouth
- ✓ Crap shape mouth
- ✓ Café au lait macules
- ✓ 5th finger Clinodactyly
- ✓ Syndactyly
- ✓ Excessive sweating
- ✓ Male genital abnormalities
- ✓ High-pitched voice
- ✓ Shoulder dimples
- ✓ Failure to thrive
- ✓ Speech delay

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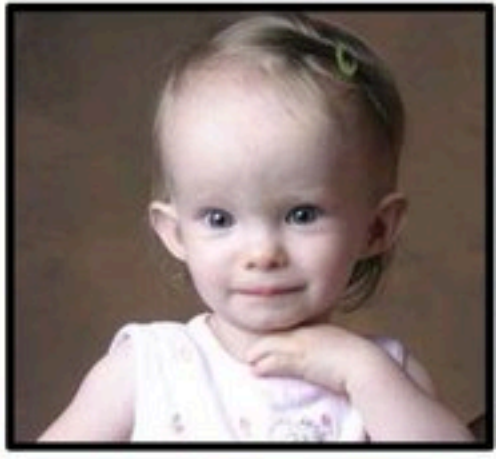
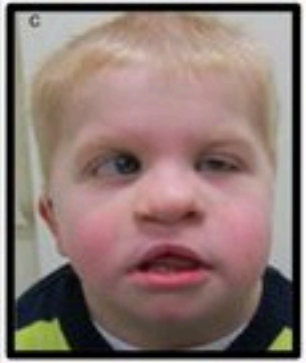
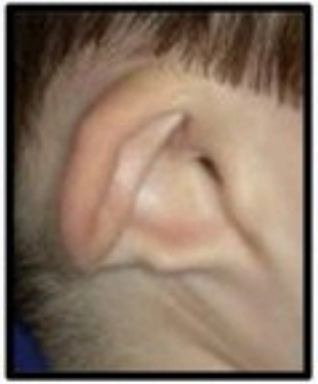
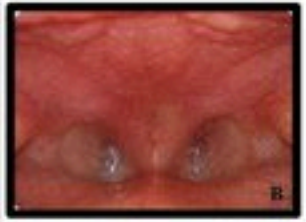
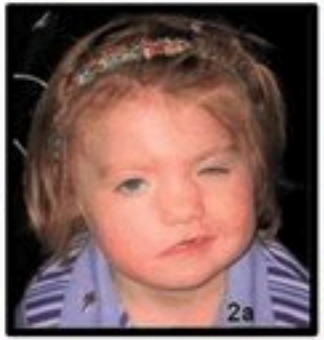
❖ Mode of inheritance?

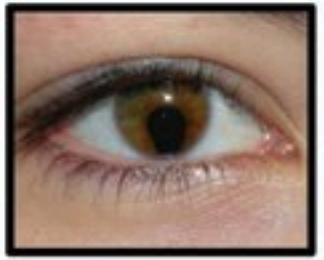
- ✓ Hypomethylation of IGF-2, or
- ✓ Maternal UPD of chromosome 7

❖ Laboratory finding ?

- ✓ Hypoglycemia



43) CHARGE Syndrome

❖ Diagnostic criteria

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Major criteria

- ✓ Ocular colobomas
- ✓ Choanal atresia
- ✓ Cranial nerve dysfunction (Anosmia, facial palsy, auditory nerve hypoplasia)
- ✓ Ear anomalies

Minor criteria

- ✓ Genital hypoplasia
- ✓ Developmental delay
- ✓ Cardiovascular anomalies
- ✓ Cleft lip and/or palate
- ✓ Tracheoesophageal fistula or esophageal atresia
- ✓ Facial dysmorphology (Square face, prominent forehead, flat midface)
- ✓ Growth failure
- ✓ 'J'-shaped hockey stick palmar crease

Definite CHARGE syndrome:
- ✓ 4 major Or
- ✓ 3 major and 3 minor

Probable CHARGE syndrome:
- ✓ 1 or 2 major and several minor.

❖ Mode of inheritance?

- ✓ Autosomal dominant
- ✓ CHD7 gene

❖ What is the most common cardiac defect?

- ✓ Tetralogy of fallot
- ✓ Conotruncal defect
- ✓ Arch anomalies

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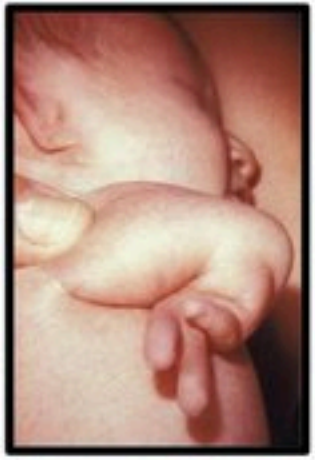

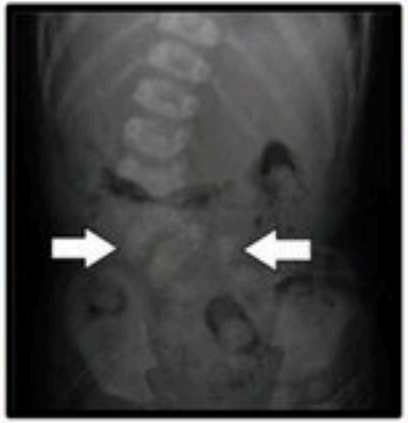

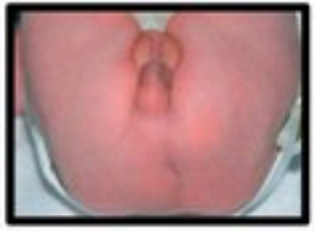

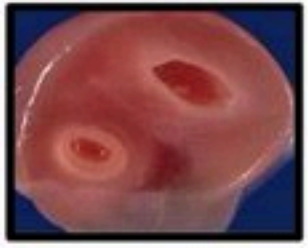

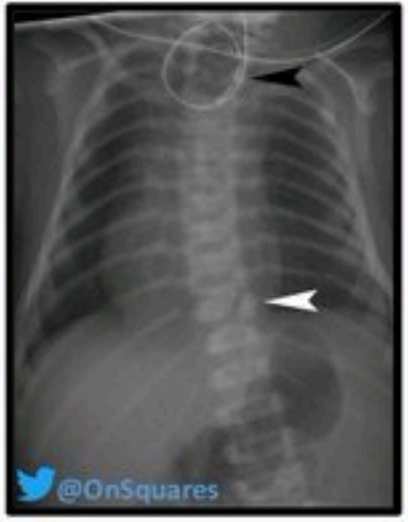

44) VACTERL association

❖ Clinical features

- ✓ Vertebral defect
- ✓ Anal atresia
- ✓ Congenital heart defect
- ✓ Tracheoesophageal fistula
- ✓ Esophageal Atresia
- ✓ Renal anomalies
- ✓ Limb (radial) hypoplasia

±

- ✓ Single umbilical artery

❖ How to confirm the diagnosis?

- ✓ Clinically
- ✓ ≥ 3 features

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❖ Mode of inheritance?

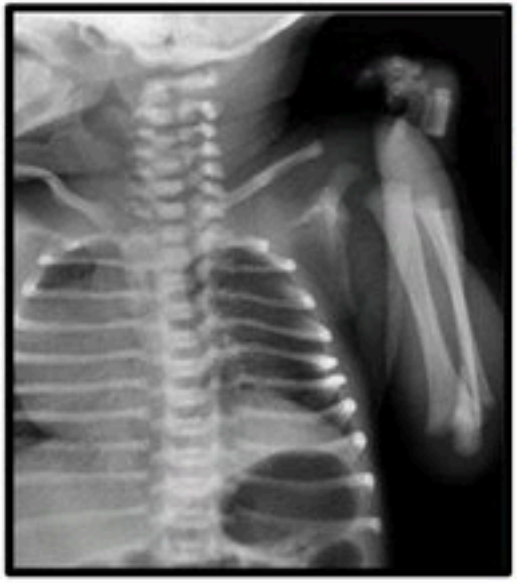
- ✓ Sporadic

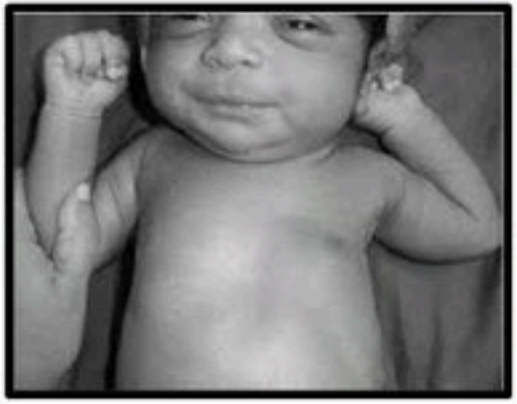
❖ What is the most common cardiac defect?

- ✓ **VSD**
- ✓ TOF
- ✓ ASD
- ✓ PDA

❖ What is the level of intelligence ?

- ✓ Normal
- ▪ Note CHARGE have intellectual disability





45) Poland Syndrome

❖ **Clinical features**

- ✓ Unilateral absence of the pectoralis muscle.
- ✓ Aplasia of ipsilateral
  - ▪ Ribs
  - ▪ Costal cartilages
  - ▪ Nipples
- ✓ Abnormalities of ipsilateral proximal & distal upper extremity
- ✓ Dextrocardia
- ✓ Diaphragmatic hernia

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❖ **Which maternal disease increase the risk of Poland syndrome?**

- ✓ Maternal diabetes.

❖ **Mode of inheritance?**

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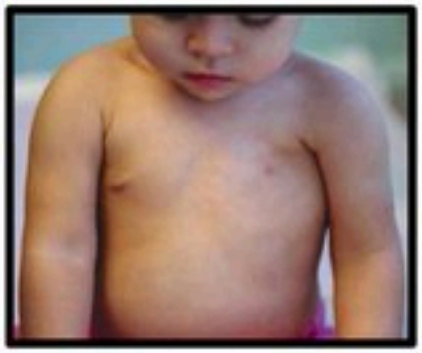
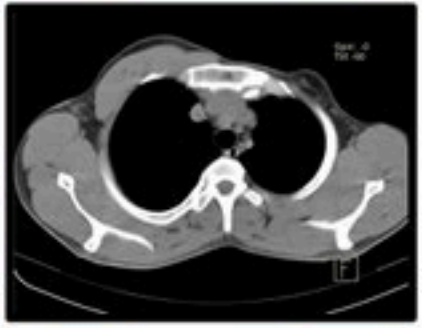
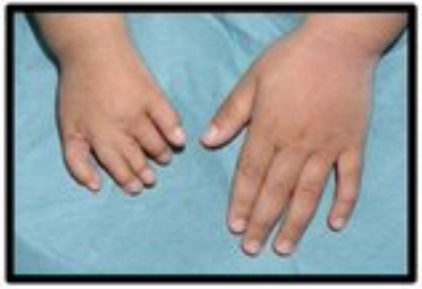
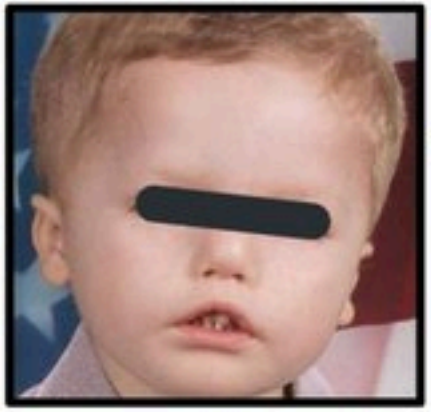
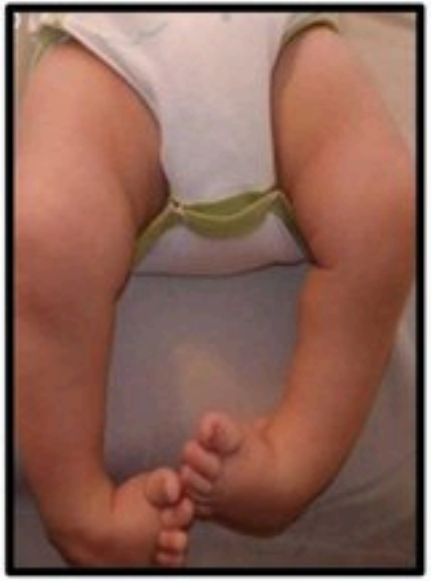
- ✓ Not inherited, unknown cause



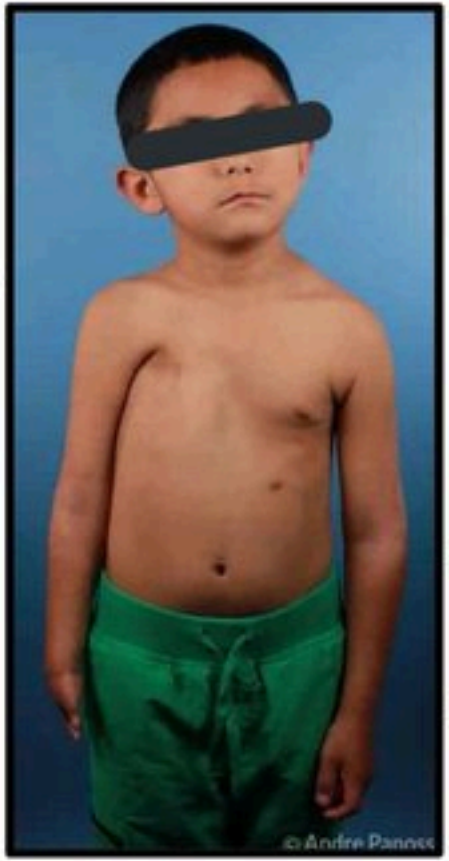








46) Möbius Syndrome



❖ **Clinical features**

- ✓ Facial (VII) nerve paralysis
- ✓ Abducens (VI) nerve paralysis
- ✓ Expressionless
- ✓ Strabismus
- ✓ Club feet
- ✓ Absent fingers or toes
- ✓ Unilateral chest deformity
- ✓ Tongue atrophy
- ✓ Dental problems

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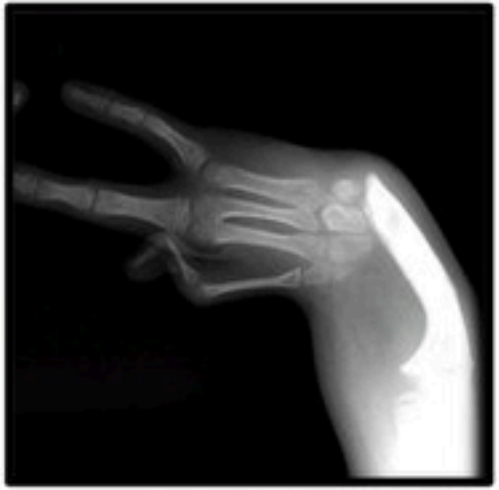
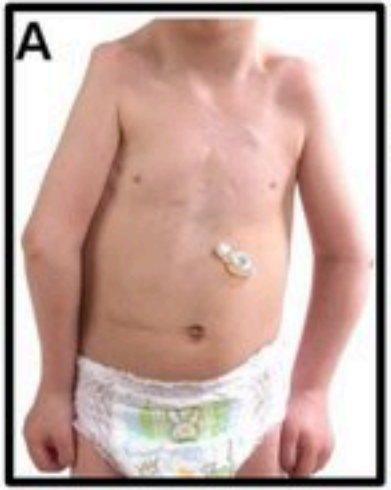
❖ **Mode of inheritance?**

- ✓ Autosomal dominant

❖ **Strong association with which syndrome ?**

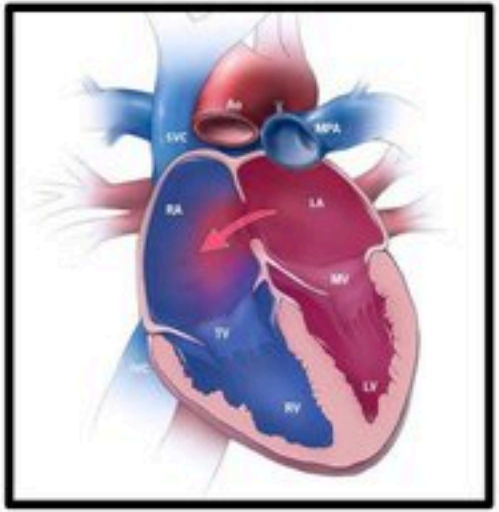
- ✓ Poland Syndrome

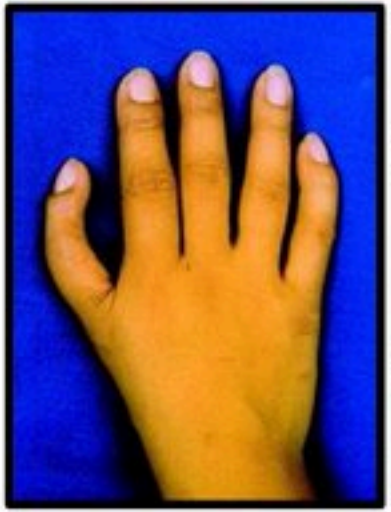




47) Holt Oram Syndrome

- **Clinical features**
  - ✓ Thumb
    - Aplasia / hypoplasia
    - Triphalangeal
  - ✓ Radial hypoplasia
  - ✓ Clavicle hypoplasia



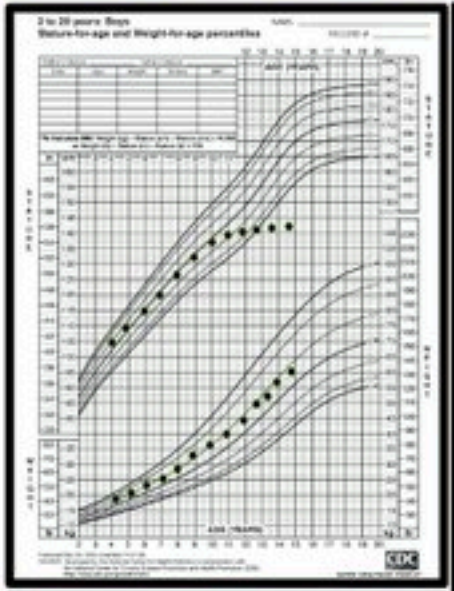


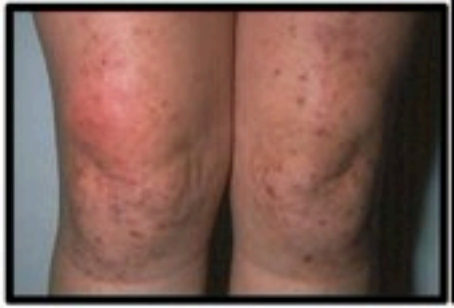
- **Mode of inheritance?**
  - ✓ Autosomal dominant

- **What is the most common cardiac defect?**
  - ✓ Atrial septal defect (ASD)
  - ✓ 1st degree heart block

NO hematological abnormalities

Celiac Disease "Gluten Sensitive Enteropathy"





Definition:

- ✓ Is an autoimmune enteropathy characterized by chronic inflammation of the small intestinal mucosa triggered by gluten uptake in genetically susceptible individuals.

At which part of bowel the inflammation of celiac disease starts?

- ✓ Proximal small intestine (duodenum) and progresses distally

What is the MOST common extra-intestinal manifestation?

- ✓ Iron deficiency anemia, unresponsive to iron therapy (Why?)

What is the pathognomonic dermatological feature ?

- ✓ Dermatitis herpetiformis

Which syndromes are at higher risk for celiac disease?

1. Down syndrome
2. William syndrome
3. Turner Syndrome

- Also Celiac disease can associated with other autoimmune disease

Which food items contains gluten ?

- ✓ Wheat, rye, barley & oats

Which enzyme is responsible for the deamidation of glutamine residues in gluten, thus triggering the cascade that results in villous atrophy?

- ✓ Tissue transglutaminase

Diagnosis

What is the initial recommended serum test?

- ✓ Tissue transglutaminase (tTG) antibody IgA

What is the most sensitive and specific antibody test ?

- ✓ IgA - Tissue transglutaminase (tTG) antibody

Followed by :

- ✓ IgA - Anti-endomysial antibody (EMA)
- ✓ IgA - Anti-gliadin antibody (AGA)
- ✓ IgA Anti-deamidated gliadin peptide antibody (DGP)
- ✓ IgG Anti-deamidated gliadin peptide antibody (DGP)

All antibodies test Should be done while the patient on gluten containing diet

Which laboratory findings can cause a false-negative screen for anti-tTG IgAs?

- ✓ Low or absent levels of IgA

What is the gold standard test for diagnosis?

- ✓ Small Intestine biopsy (before start gluten free diet)

What is the classic histological finding?

- ✓ Increase intraepithelial lymphocytes (>25 IEL/100 enterocytes)
- ✓ Crypt elongation
- ✓ Partial or total villous atrophy

What human leukocyte antigen (HLA) type is commonly associated with Celiac disease?

- ✓ HLA-DR3-DQ2
- ✓ HLA-DR4-DQ8.

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Treatment & Follow Up

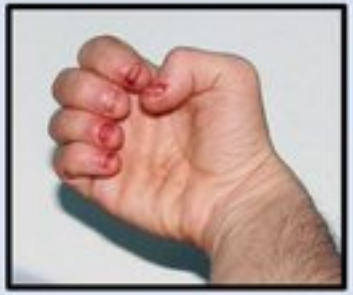
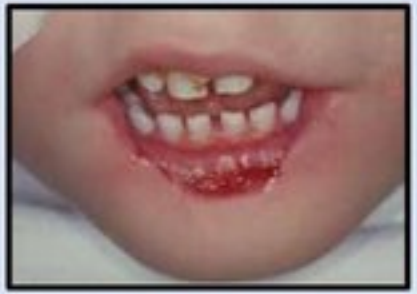
What is the main treatment of celiac disease?

- ✓ Gluten free diet

How to follow up the compliance with dietary therapy?

- ✓ Normal tTG IgA levels (Zero or low level)

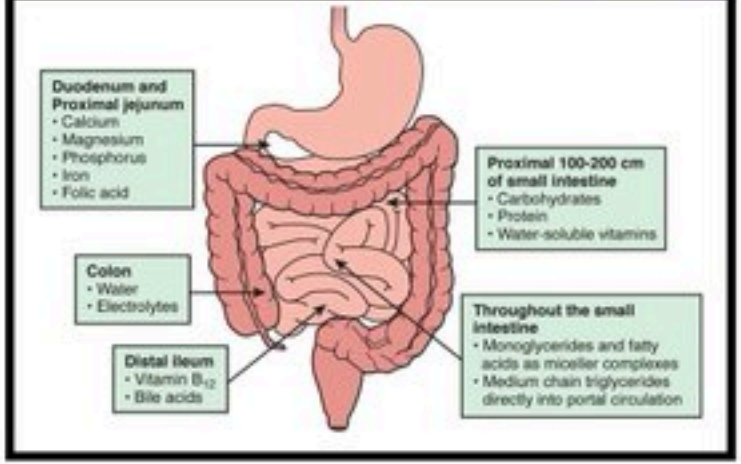
Lesch Nyhan Disease


- It is a disorder of purine metabolism due to **HPRT** (Hypoxanthin-Guanine Phosphoribosyltransferase) deficiency.
- Caused by mutations in the **HPRT1 gene** and is inherited as an **X-linked recessive**.
- Characterized by **neurological** and **behavioral** abnormalities and **hyperuricemia**.
- Neurological abnormalities include **dystonia, chorea, hypotonia and ballismus**, also they are usually cannot walk.
- **Self-injury** (including fingers and mouth biting and head banging) is the most common and distinctive behavioral problem, also they have **developmental delay**.
- **Hyperuricemia** can cause **gouty arthritis**, kidney and bladder **stones**.
- Treatment is symptomatic and supportive focuses on prevention of renal failure by pharmacologic treatment of hyperuricemia, with high fluid intake along with alkalization and allopurinol.
- Affected people often do not survive past the first or second decade of life due to renal failure.

Short bowel syndrome

- At **birth**, the length of small bowel is 200-250 cm; by **adulthood**, it grows to 300-800 cm.
- Why bowel resection in an infant has a better prognosis than in an adult ?
  - ✓ Because of the potential for intestinal growth and adaptation.



- How much bowel do infant need to survive and be eventually weaned from parenteral nutrition?
  - ✓ 15 cm of bowel with an ileocecal valve
  - ✓ 20 cm without an ileocecal

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Table 364.11	Causes of Short Bowel Syndrome
CONGENITAL	
Congenital short bowel syndrome	
Intestinal atresia	
Gastroschisis	
BOWEL RESECTION	
Necrotizing enterocolitis	
Volvulus with or without malrotation	
Long segment Hirschsprung disease	
Meconium peritonitis	
Crohn disease	
Trauma	