PREFACE

"Do not forget to ask yourself why"

Prof. Khalid Naji (1923 - 2008)

This book (Iraqi Color atlas Of Pediatrics: Questions& Answers) is a short pediatric case files in the form of picture test, the pictures are of the most common pediatric problems that we used to see in our practice in this country. The idea behind writing this book is that our students, residents and young colleagues are familiar with similar books from other parts of the world but not from their area, and so they will find it helpful and entertaining. Our interest in medical education and the recent advances in evaluation of medical students and residents were the main inspiration for us to write this book and these cases could be used in the clinical examinations of students, residents and postgraduate colleagues. Our knowledge and experience had increased as we learned a lot from our very nice and cooperative patients and their families.

This book consist of two parts, the first is for the pictures of patients and the questions, the second part is for answers, explanations and suggested references. The best way of reading this book, is to read the questions in groups, to answer these questions and then

compare your results with the correct answers in the 2nd part of the book and to have a look on the details of the explanations.

We would like to express our gratitude to our patients and their families for they made it worthwhile. We are grateful to our students and residents for their stimulation in writing this book. Our thanks should also be advanced to our colleagues in the department of pediatrics for their continuous suggestions. we are most appreciative of the efforts of Mr. Baker M. Adnan ,BSC computer sciences and Mr. Ehab Jaber, who reviewed the electronic copy of this book and gave many constructive suggestion.

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A'alan H Al-Zamili

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2014

Figure 1

Q. What is the recommended investigation in this condition?

Figure 2

Q1: What are the steps in physical examination of this condition?

Q2: What are the sequale of delayed treatment?

Figure 3

Q1. Mention 2 signs in this child?

Q2. What are their causes?

Figure 4

Q1. What is the term given to this abnormality?

Q2. What are the possible underlying causes?

Figure 5

Q1. Mention 3 complications of this condition?

Q2. Is there an indication for specific treatment?

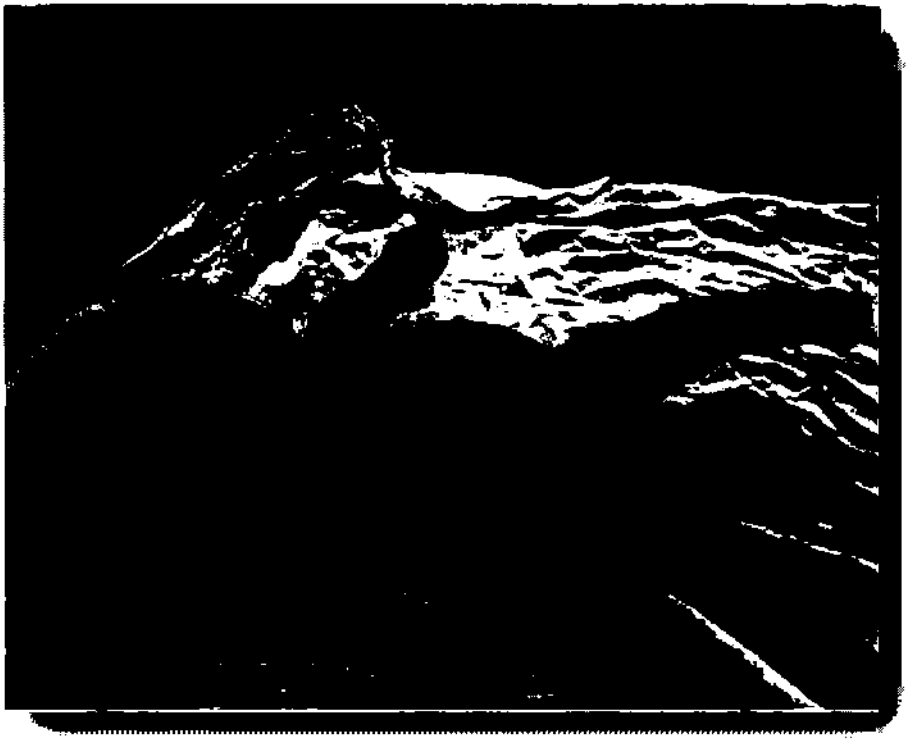


Figure 6

Q. What is the pathophysiology of this condition?

Figure 7

Q1. What is the bed side test to document the diagnosis?

Q2. What is the treatment?

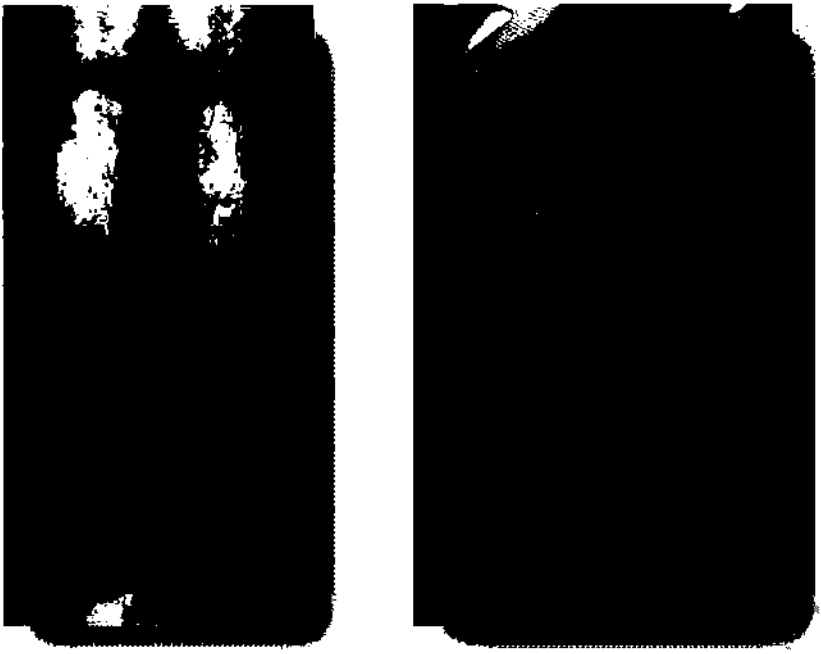


Figure 8

Q. What are the indications of steroid in this condition?

Figure 9

Q. Mention 3 physical signs in this neonate?

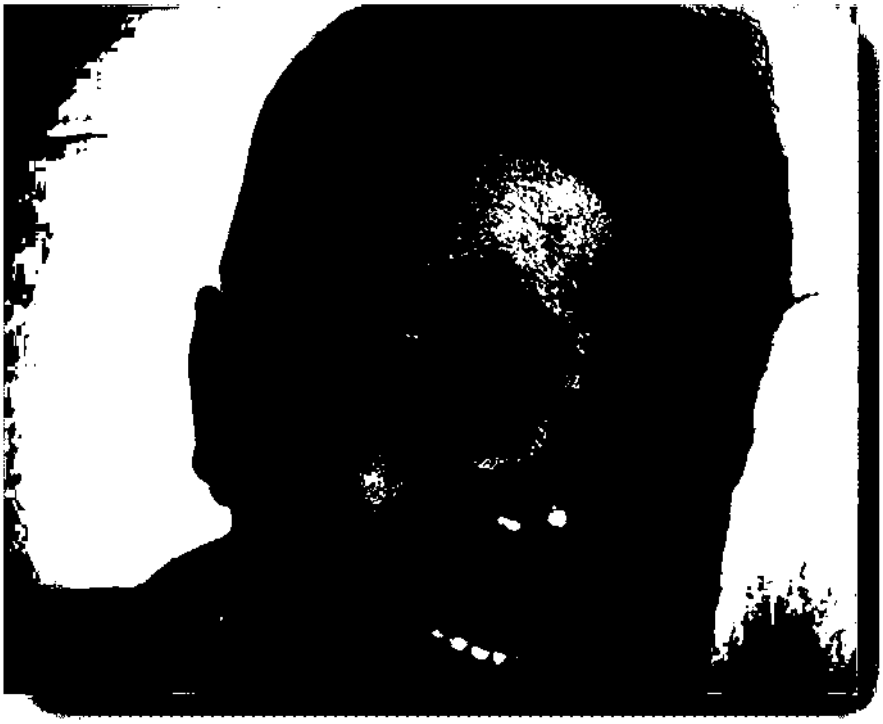


Figure 10

Q1. What is the name given to this sign?

Q2. Mention 4 possible causes?

Figure 11

Q1. What are the other sites that should be examined?

Q2. Mention 3 complications of this disease?

Figure 12

Q. What are the complications of this treatment?

Figure 13

Q. What is the term given to this face?

Figure 14

Q1: Mention 2 signs in this infant?

Q2: Mention 2 syndromes associated with these signs?

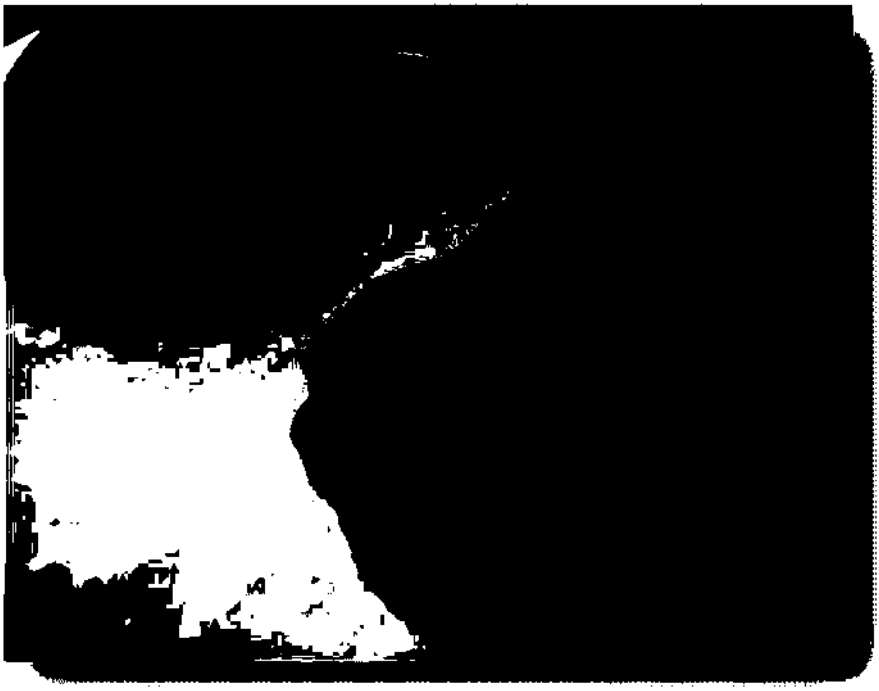


Figure 15

Q. What are the classical causes of this condition?

Figure 16

Q1. Is there a need for treatment?

Q2. What is the underlying cause?

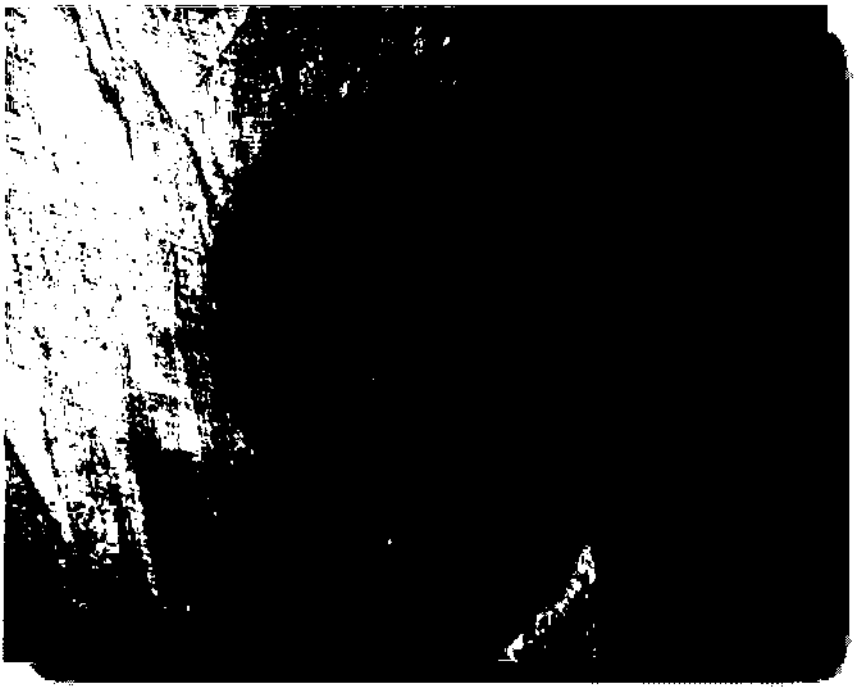


Figure 17

Q1. What are the effects of this condition?

Q2. What other diagnosis that should be excluded?

Figure 18

Q1. What is the diagnosis?

Q2. What is the pathophysiology?

Figure 19

Q1. When the diagnosis should made?

Q2. What are the things that should be assessed in the follow up of this baby?

Figure 20

Q1: what is the type of this disease?

Q2: where it should be treated?

Figure 21

Q. What are the other possible associated anomalies with this condition?

Figure 22

Q: what are the hematological sequale of this condition?

Figure 23

Q1. What is the appropriate time for surgery?

Q2. What are the possible complications?

Figure 24

Q. What are the steps of management?

Figure 25

Q. Mention 4 possible causes?

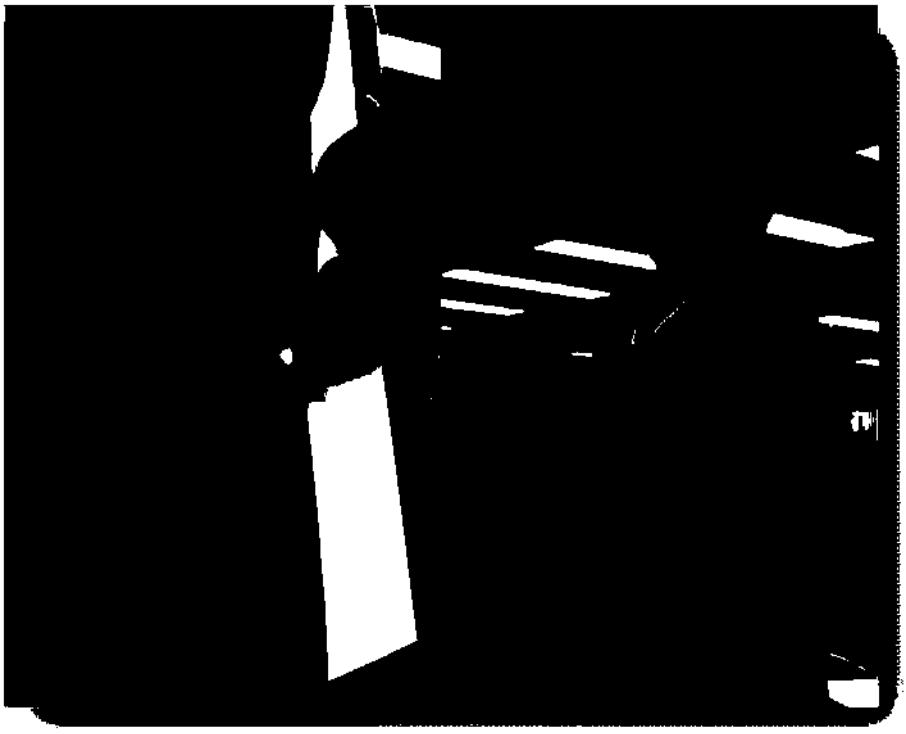


Figure 26

An8 years old boy with jaundice, ascites and hepatomegaly with positive family history

Q1: What are the recommended investigations?
Q2: What are the lines of treatment?

Figure 27

Q1. What abnormality is seen in the eye?

Q2. Mention three syndromes that are associated with this abnormality?

Figure 28

Q. Mention 4 diagnostic investigations in this condition?

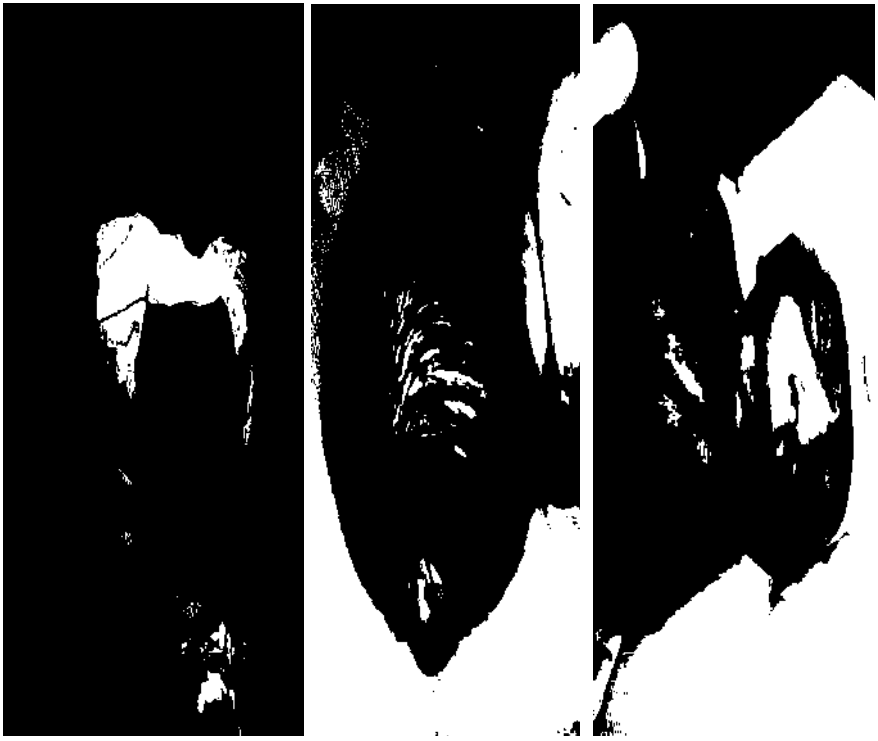


Figure 29

Q1. What are the possible underlying causes of these conditions?

Q2. What is the embryological basis?

Figure 30

Q1: What are the screening tests for this condition?

Q2: What are the late complications of this disease?

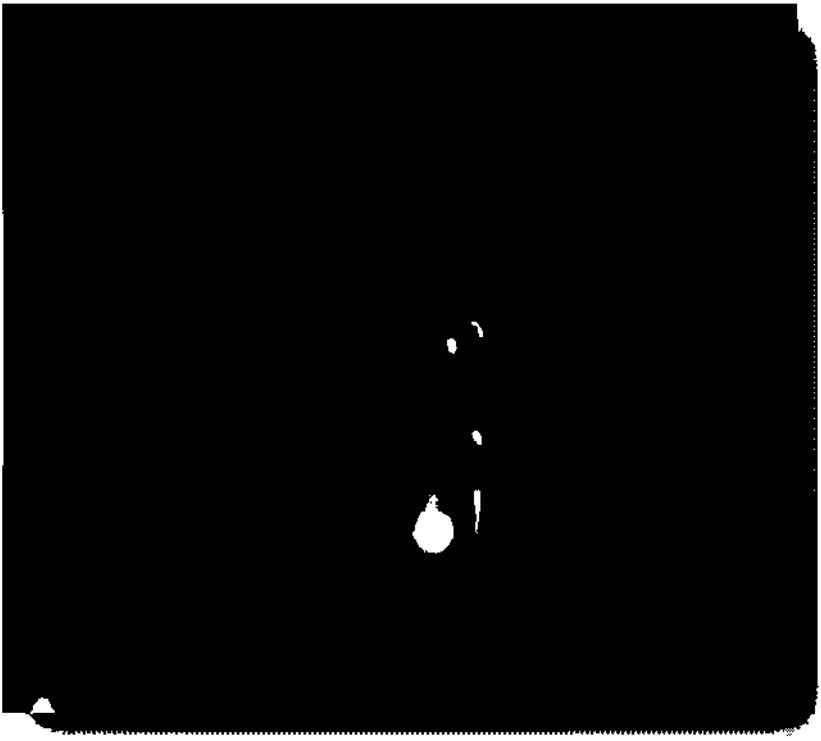


Figure 31

Q. What is abnormal in this child?

Figure 32

Q1. What is the chromosomal basis of this condition?

Q2. What are the endocrine diseases that are associated with this condition?

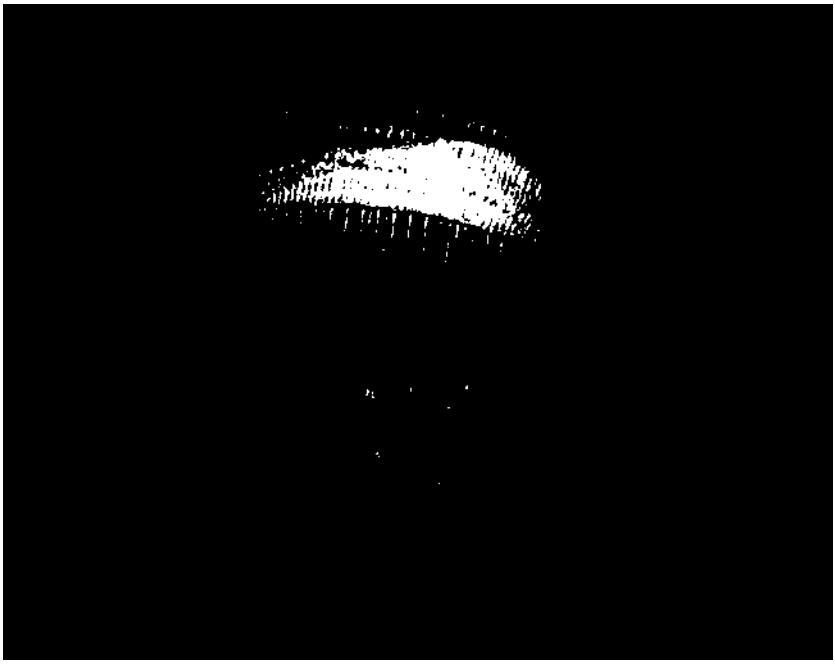


Figure 33

Q1. Mention 3 signs in this infant?

Q2. How to confirm the diagnosis?

Figure 34

Q1. What is the name that is given to this lethal anomaly?

Q2. What is the embryological basis of this condition?

Figure 35

Q1: Which one should be operated first?

Q2: Mention 3 syndromes that are possibly associated with this condition

Figure 36

Q1: What is abnormal in this infant?

Q2: Mention 3 possible associated diseases.

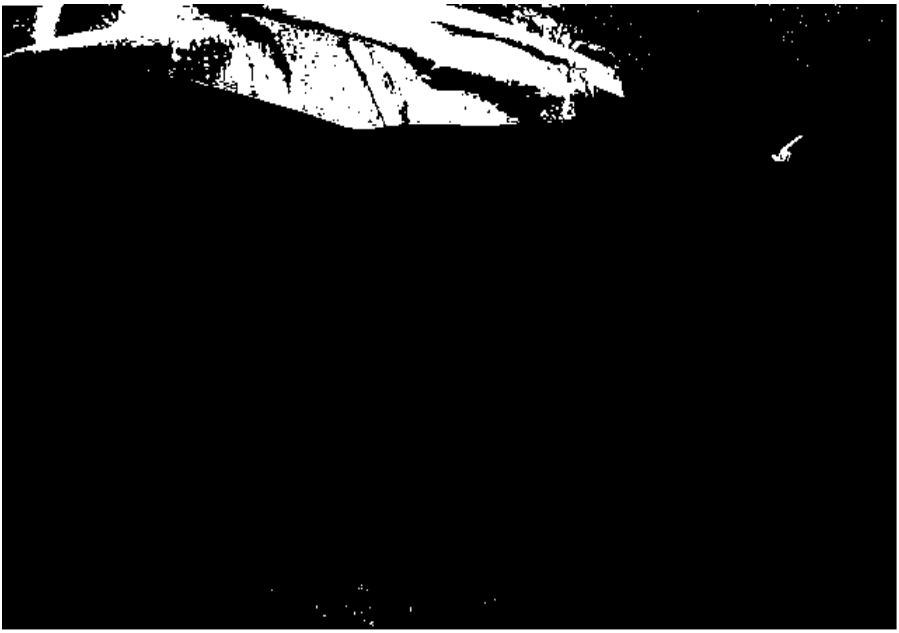


Figure 37

Q1. Is this condition need treatment?

Q2. What is the pathophysiology behind it?

Figure 38

Q1. What is the type of this rash?

Q2. Mention 4 complications of this condition?

Figure 39

Q1. Is this a usual site?

Q2. What is the treatment?

Figure 40

Q1. What is the diagnosis?

Q2. What is the treatment?

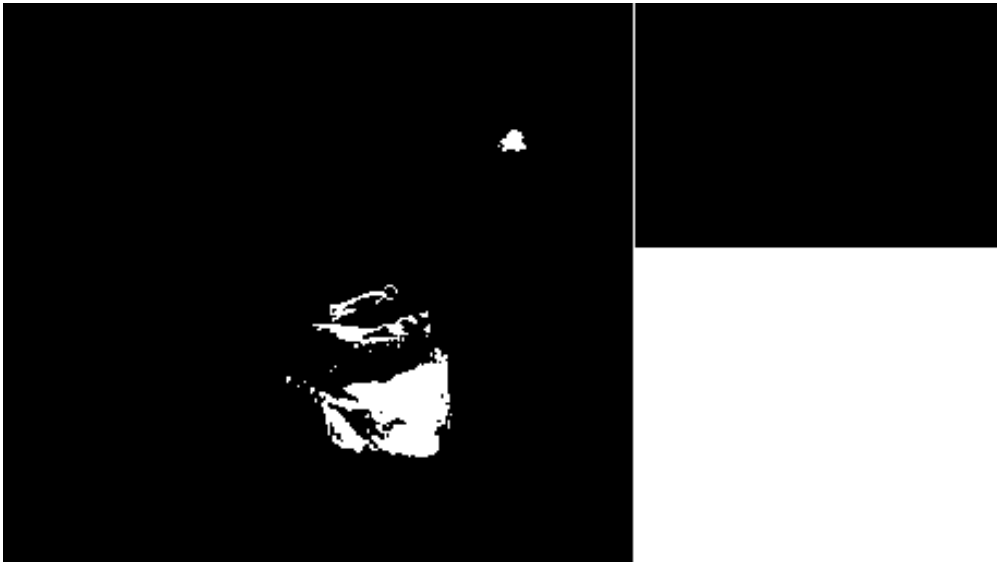


Figure 41

Q. Mentions three problems that this neonate has?

Figure 42

Q1. What is the diagnosis?

Q2. What is the pathophysiology?

Figure 43

Q1. What is the diagnosis?

Q2. Mention three differential diagnosis?

Figure 44

Q1. What is the pathophysiology?

Q2. What is the risk for chromosomal abnormalities?

Figure 45

This infant had recurrent chest infection & chronic diarrhea

Q1. What is the most likely diagnosis?

Q2. How to confirm the diagnosis?

Figure 46

Q1. Mention two physical signs in this girl?

Q2. What are the possible diagnoses?

Figure 47

Q. What are the new treatments of this condition?

Figure 48

Q. What are the three signs that this neonate had?

Figure 49

Q1.What is the pathophysiology of this condition?

Q2.What is the treatment?

Figure 50

Q1.What is the most likely diagnosis?

Q2.What is the treatment?

Figure 51

Q1.What is wrong in this neonate face?

Q2.What is the prognosis?

Figure 52

This neonate had 4days history of fever & irritability before the onset of this rash.

Q1.What is the causative organism?

Q2.What are the possible complications of this disease?

Figure 53

Q1. What is the diagnosis?

Q2. Which one is more liable for complication?

Figure 54

Q1. What is the diagnosis?

Q2. What is the prognosis?

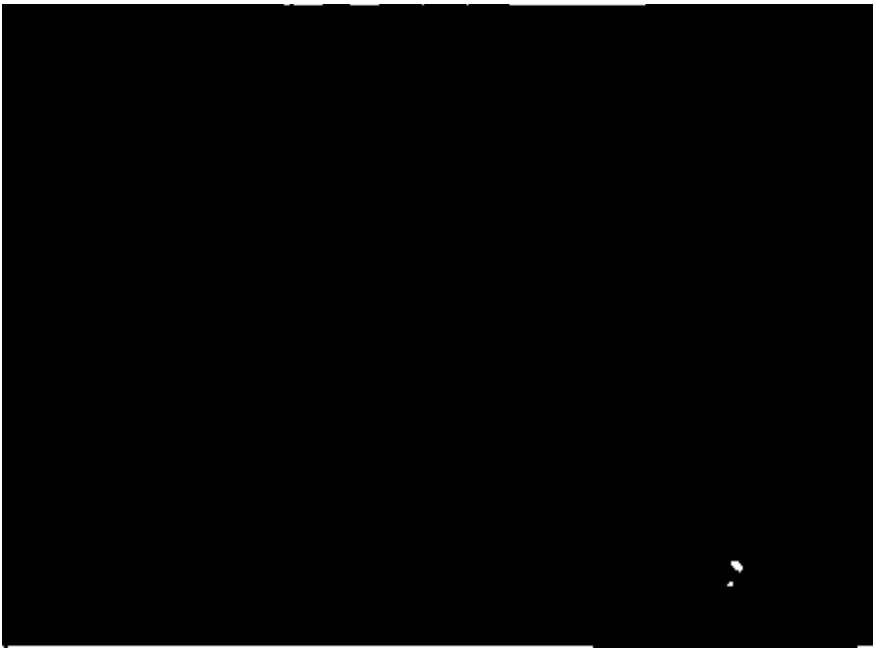


Figure 55

Q1. What is the abnormality in the eye?

Q2. What are the possible causes?

Figure 56

Q1. What are the most likely diagnosis?

Q2. What are recommended investigations?

Figure 57

Q1. Mention 3 possible diagnosis.

Q2. What are the recommended investigations and treatment?

Figure 58

Q1. Mention 4 abnormalities that this baby is having.

Q2. What are the most common problems that he is liable for?

Figure 59

Q. What is the diagnosis?

Figure 60

Q. What is the treatment of this condition?

Figure 61

Q.1.What is the diagnosis?

Q.2.What surgical operation was done for this infant?

Figure 62

Q. What is the diagnosis?

Figure 63

Q. Mention 4 possible differential diagnoses?

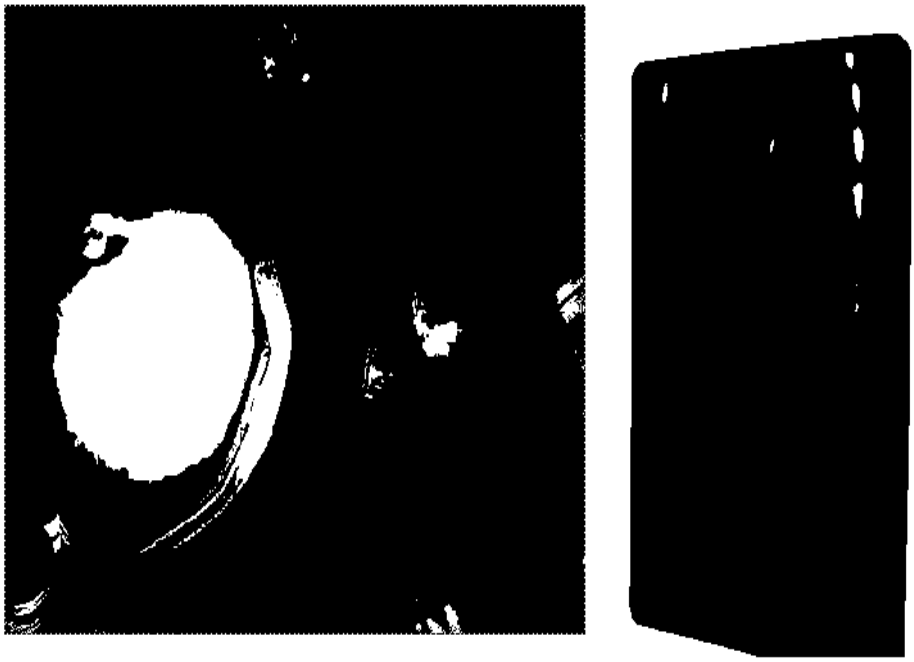


Figure 64

Q. What are the possible differential diagnoses in this child?

Figure 65

Q. What are the conditions that are associated with this sign?

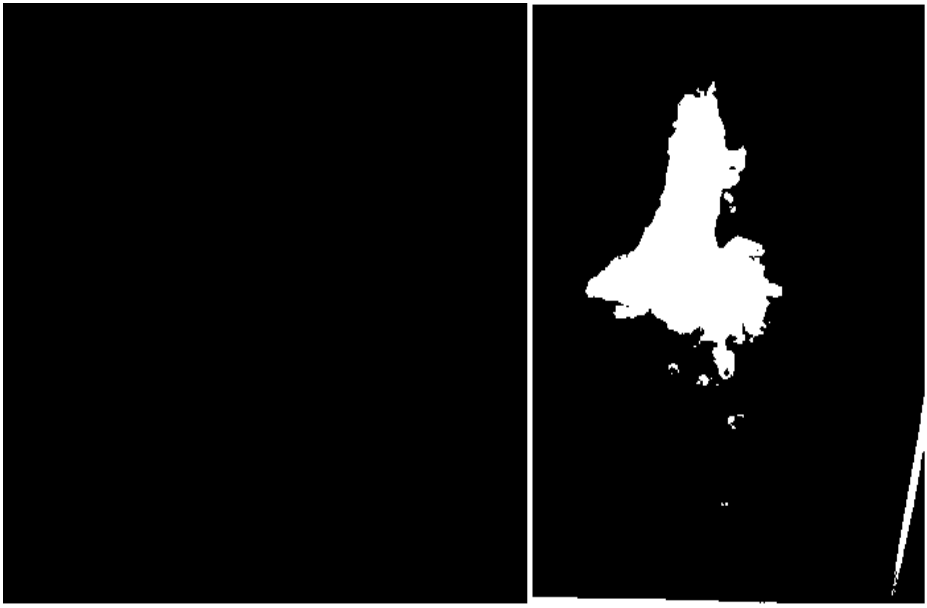


Figure 66

Q. What is the diagnosis?

Figure 67

Q. What is abnormal in this infant?

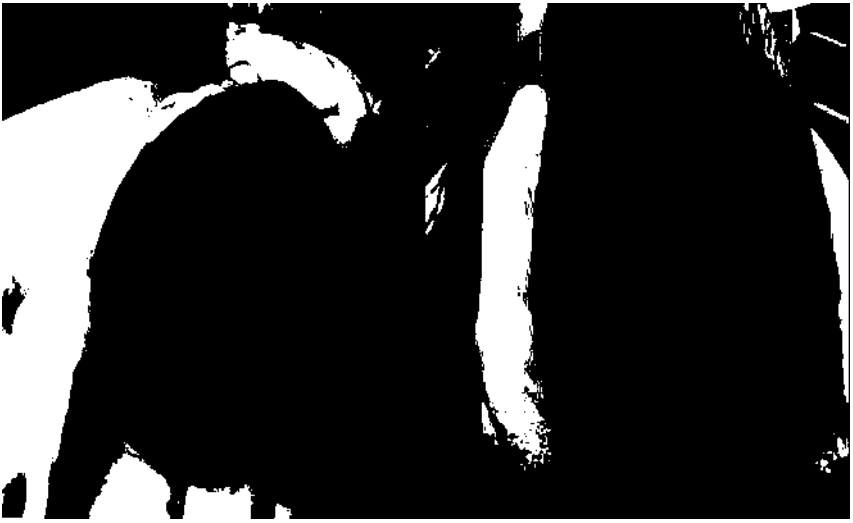


Figure 68

Q. What is the diagnosis? What is the treatment?

Figure 69

Q. What are the X- ray findings in this disease?

Figure 70

Q. Mention 2 signs in this neonate, what are the predisposing factors?

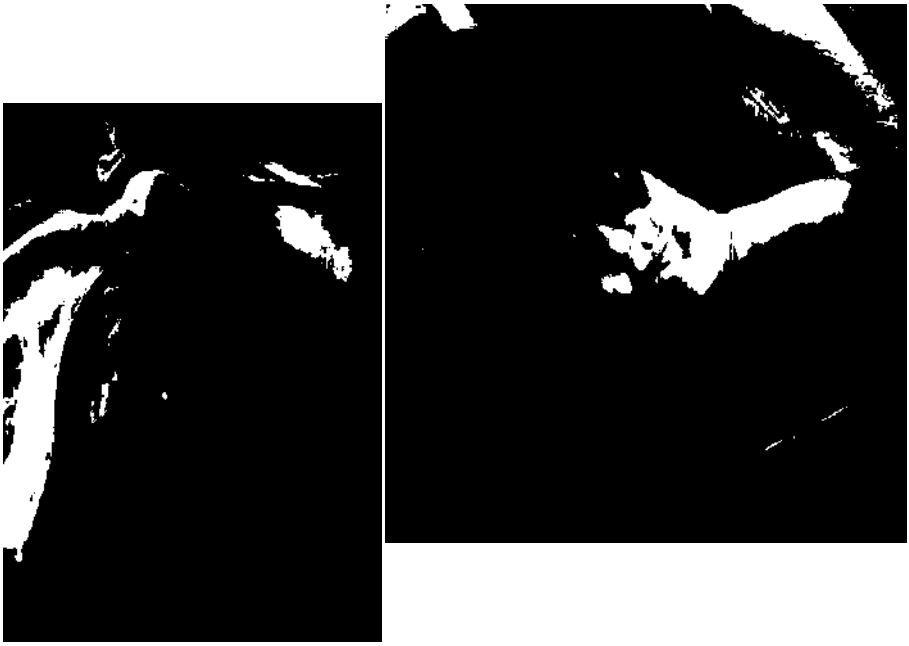


Figure 71

Q. Mention 4 possible diagnoses?

Figure 72

Q. What are the new lines of treatment of this condition?

Figure 73

Q. What is the diagnosis? What are the predisposing factors?

Figure 74

Q. Mention 4 possible causes for this condition in the neonate?

Figure 75

Q. Mention 2 signs in this patient, what is the most likely cause?

Figure 76

Q1. What is the diagnosis?

Q2. What are the types and recent therapies of this disease?

Figure 77

Q1. What is the diagnosis?

Q2. What is the explanation for the occurrence of this sign?

Figure 78

Q1. What is the diagnosis?

Q2. What are the associated conditions with this syndrome?

Figure 79

Q1. What is the diagnosis?

Q2. What are the lines of treatment?

Figure 80

A 3 -year o l d boy presented with recurrent epistaxis and echymosis since early infancy.

Q1. What is the diagnosis?

Q2. What are the suggested investigations?

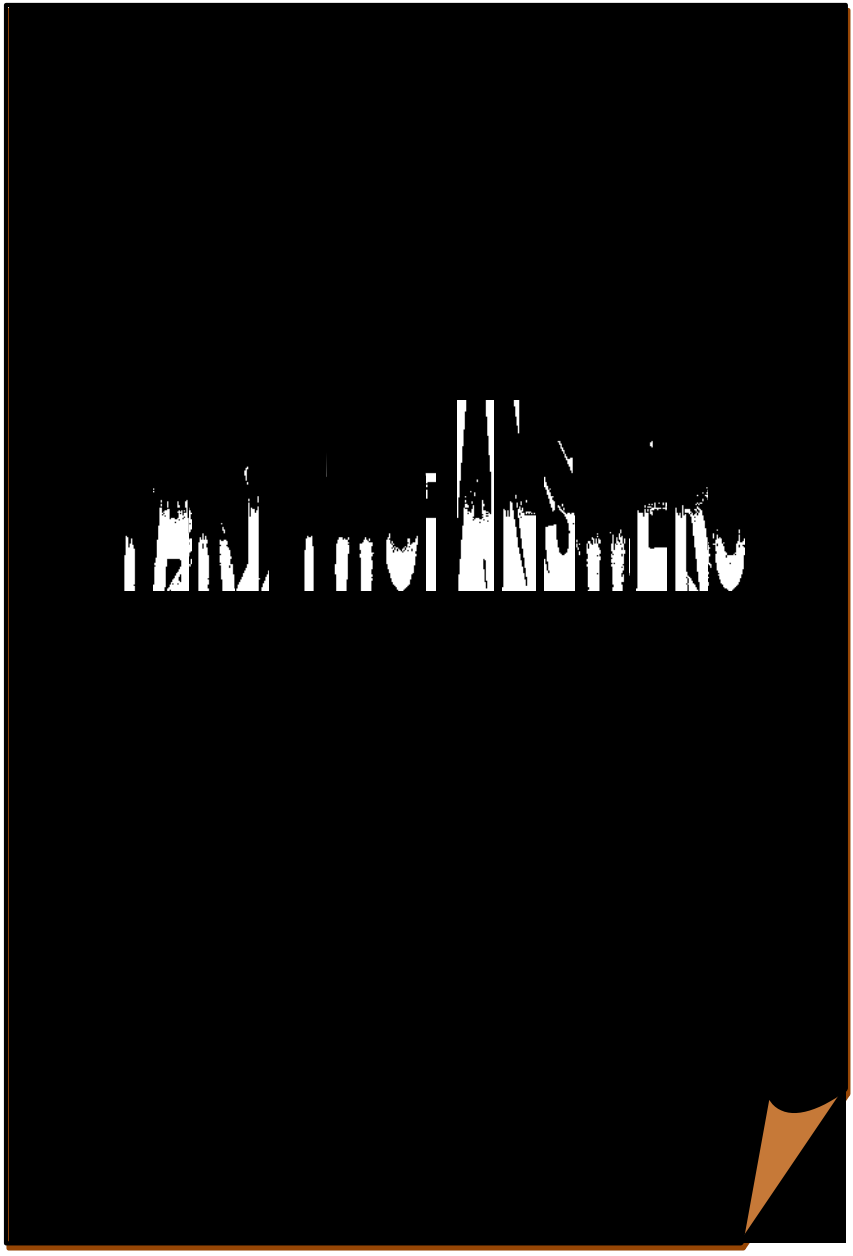
Figure 81

Q. What is your diagnosis?

Figure 82

Q1: what is the diagnosis?

Q2: what is the treatment?



Answers

1. The recommended investigations in ITP(idiopathic thrombocytopenic purpura) include:

a. Hemoglobin value, white blood cells (WBC) count , differential count, platelets count (low , the PLT size is normal or increased)

b. Indications for bone marrow aspiration include: an abnormal WBC count or differential, unexplained anemia, findings suggestive of bone marrow disease on history and physical examination

c. Other lab tests: indicated by the history and physical examination (Antinuclear antibody test in an adolescent with new onset ITP, HIV studies in at risk patients, Coombs test in unexplained anemia to rule out Evans syndrome).(1)

2. Strabismus

Strabismus is the misalignment of the eyes with either an in-turning (esotropia), out-turning (exotropia), or up-turning(hypertropia) of one eye.

1. **a.** assessment of the visual acuity

b. corneal light reflex tests (Hirschberg test, the Krimsky method using prisms)

c. the cover tests: requires child attention and cooperation, good eye movement capability, and reasonably good vision in each eye. They consist of the cover-uncover test and the alternate cover test

2. Amblyopia and devastating vision loss that might occur even with strabismic deviation too small to be evident by gross inspection (2)

3.

1. a. slit like umbilicus (edema of the abdominal wall)
   b. erythema and purple striae
2. nephrotic syndrome, steroid therapy

Striae cutis distenae are bands of atrophic skin occur most frequently in areas that have been subject to distension. The most common causes are rapid growth, pregnancy, obesity, cushing disease, prolonged corticosteroid therapy (3).

4.

Persistent fisting of hands after 3 months of age – suggestive of hypertonia/ cortical spinal tract dysfunction (4).

5.

1. Varicella (chicken pox) is the manifestation of the primary infection caused by Varicella-zostervirus, the complications of this disease include : secondary bacterial infection of the skin , encephalitis (1 / 50000 cases and cerebellar ataxia (1/ 4000 cases),pneumonia, thrombocytopenia, arthritis,myocarditis, pericarditis, hemolytic -uremic syndrome. (5)

2. Acyclovir therapy is not routinely recommended for the treatment of uncomplicated varicella in healthy child , oral therapy (20 mg/kg/dose maximum 800 mg/dose

given as 4 doses for 5 days is used to treat uncomplicated varicella in children more than 13 years and children more than 12 months of age with: chronic cutaneous or pulmonary disorder; receiving short term, intermittent or aerosolized corticosteroids; receiving long-term salicylate therapy; and possibly second cases in household contacts. Treatment should be initiated within 24 hr of the onset of the exanthema.

Intravenous therapy is indicated for severe disseminated (pneumonia, severe hepatitis, thrombocytopenia or encephalitis) disease and in immunocompromised patients 500 mg/m2 every 8 hours within 72 hours of initial symptoms for 7 days.(6)

6.

Amniotic band syndrome (ABS):

There are several theories as to the cause of ABS. The most widely accepted is a rupture of the amnion occurring early in gestation. The fibrous bands of amnion that occur from the amniotic rupture encircle the limbs, resulting in tourniquet-like defects and intrauterine amputations. The timing of the rupture is believed to occur between 28 days after conception to 18 weeks of gestation. However, late bands can occur and present at birth with multiple abnormalities of the limbs, even after a normal sonogram was performed earlier in gestation. This can be observed following any form of intervention such as amniocentesis or fetal surgery. (7)

7.

1. Electrocardiogram
2. IV calcium is given as bolus 20 mg/kg CaCl2, or 100 mg/kg calcium gluconate at a rate of 0.5-1 ml/minute while the heart rate is monitored; some patients require a continuous IV calcium drip.

   Prolonged Q-Tc interval more than 0.45 seconds is a feature of hypokalemia, hypocalcaemia and long Q-T syndromes, in hypokalemia a U wave may be noted at the end of the T wave.

   The Q-Tc can be calculated by dividing the measured Q-T interval by the square root of the preceding R-R interval. (8)

8.

The indications of corticosteroids in Henoch-Schonlein purpura are:

a. Intestinal complications (hemorrhage, obstruction, and intussusception) in addition to other required treatment

b. CNS complications

c. rarely in chronic or recurrent HSP which may respond to pulse doses of IV methyprednisolone.(9)

9.

This neonate had: jaundice, subconjunctival hemorrhage and ophthalmia neonatorum

Ophthalmia neonatorum is conjunctivitis occurring in infants younger than 4 weeks of age. The classical causes are : Silver nitrate instillation, Neisseria gonorrhoeae, Pseudomonas aeruginosa, Chlamydia trachomatis and Staph aureus . Treatment of gonoccocal ophthalmia is by ceftriaxone 50mg /kg/day for 1 dose , not to exceed 125 mg ; irrigation of the eye with saline every 10-30 minutes gradually increasing to 2-hr intervals until the purulent discharge has cleared. Treatment of inclusion blennorrhea (Chlamydia) is with oral erythromycin 50 mg/kg/day in 4 divided doses. Pseudomonas conjunctivitis is treated with systemic antibiotics , including an aminoglycoside, plus local saline irrigation and gentamycin ophthalmic ointment. Staphylococcal conjunctivitis is treated with parenteral methicillin and local saline irrigation.(10).

10.

1. Photophobia

Photophobia is eye discomfort in bright light .Severe photophobia may be associated with eye problems and cause severe eye pain even in relatively low light, or secondary to systemic diseases. Photophobia is a fairly common symptom.

2.Causes:

- Excessive wearing of contact lenses, or wearing badly fitted contact lenses
- Eye disease, injury, or infection (such as chalazion, episcleritis, glaucoma)

- Burns to the eye
- Common migraine headache
- Meningitis
- Acute iritis or uveitis
- Corneal abrasion
- Corneal ulcer
- Drugs such as amphetamines, atropine, cocaine, cyclopentolate, idoxuridine, phenylephrine, scopolamine, trifluridine, tropicamide, and vidarabine
- Eye testing in which the eyes have been dilated.(11)

11.

Scrotal edema and bilateral hydrocele

1.the other sites that should be examined are the face, legs and feet, abdomen (for evidence of ascites), chest (for evidence of pleural effusion) and evidence of sacral edema.

Nephrotic syndrome is primarily a pediatric disorder and is 15 times more common in children than adults; the incidence is 2-3 /100000 children per year. The characteristic features of nephrotic syndrome are heavy proteinuria (more than 40/m2/hr), hypoalbuminemia, edema and hyperlipidemia. Renal biopsy should be considered in children with features that make minimal change nephrotic syndrome less likely (hematuria, hypertension, renal insufficiency, hypocomplementemia, age<1 year or >8 years)

2. Complications of nephrotic syndrome include infections, thromb-oembolic events, complications of treatment(steroids: cushingoid appearance, hyper-

tension, cataracts and or growth failure; cyclophosphamide: neutropenia, dissiminated varicella, hemorrhagic cystitis, alopecia, sterility, increased risk of future malignancy; cyclosporine, tacrolimus : hypertension, nephrotoxicity, hirsutism, and gingival hyperplasia) (12).

12.

A ventriculoperitoneal shunt

The major complications of shunting are:

a. Occlusion : characterized by headache, emesis, papilledema and mental status changes

b. Bacterial infection : usually due to staphylococcus epidermidis, manifested by fever, headache and meningismus

c. Hypocomplementemic glomerulonephritis (shunt nephritis): hypertension, hematuria, anemia and elevated blood urea nitrogen. The condition is due to antigen-antibody complex deposition in the glomeruli.(13)

13.

Old man appearance :

MALNUTRITION AND SOME DEFINITIONS

Acute malnutrition

Kwashiorkor

Kwashiorkor is a severe form of *acute malnutrition.* It is characterized by clinical signs including oedema (swelling due to water retention secondary to hypoproteinemia) beginning in the lower legs and feet and which can spread to other parts of the body. Other signs include cracked and peeling skin, changes in hair color (lightening) and texture, lethargy and misery. *Marasmus*

Marasmus is a severe form of *acute malnutrition.* Marasmic individuals have the clinical signs of extreme thinness, often with flaccid skin, hanging in loose folds to give an 'old man's appearance'. Marasmic individuals may be alert but irritable.

Marasmic-kwashiorkor

Marasmic-kwashiorkor is a severe form of *acute malnutrition* when an individual shows clinical signs of both marasmus and kwashiorkor

Wasting

Wasting is a form of *acute malnutrition*. Wasted individuals are too light for their height (very thin).

Chronic malnutrition

Stunting

Stunting is a form of *chronic malnutrition* that arises when an individual is too short for their age and occurs in the first 2 to 3 years of life. Levels of stunting are likely to increase in chronic emergencies.

Underweight

Underweight individuals are too light for their age (maybe short or thin or both). (14)

14.

1.
 a. pectus excavatum
 b. micrognathia
2.
 a. Marfan syndrome
 b. Noonan syndrome

Causes of Pectus excavatum listed data base

- Aarskog syndrome
- Cardiofaciocutaneous syndrome
- Cystathionine beta-synthase deficiency
- Fragile X syndrome
- Iduronate sulphatase deficiency
- Marden-Walker syndrome
- Mucosulfatidosis
- Neurofibromatosis type 1
- Noonan's syndrome
- Oral-facial-digital syndrome type 3
- Orofaciodigital syndrome type 4
- Otopalatodigital syndrome type 1
- Prune belly syndrome
- Turner's syndrome. (15)

15.

Clubbing is a thickening of the flesh under the toe nails and fingernails. The nail curves downward, similar to the shape of the round part of an upside-down spoon. Clubbing occurs with a wide number of diseases. It is most often found in heart and lung diseases that cause a lower-than-normal amount of oxygen in the blood. Clubbing may also be due to lung cancer, and diseases of the liver and gastrointestinal tract. Clubbing may also occur in families. In this case it may not be due to an underlying disease.(16)

Causes:

Chronic lung conditions
• Bronchiectasis
• Cystic fibrosis
• Lung abscess
• Lung cancer
• Pulmonary fibrosis
Congenital heart disease (cyanotic type)
• Tetralogy of Fallot
• Total anomalous venous return
• Transposition of the great vessels
• Tricuspid atresia
• Truncus arteriosus
Digestive system diseases
• Celiac disease
• Tuberculosis of the intestines
• Cirrhosis
• Crohn's disease and ulcerative colitis
• Graves disease or hyperthyroidism
Other conditions
• Other types of cancer, including liver,
• Hodgkin's lymphoma
• Subacute endocarditis

16.

Neonatal bilateral breast hypertrophy

Breast hypertrophy related to stimulation from maternal hormones can occur in both male and female neonates

during the first few weeks of life; it is sometimes associated with a thin milky nipple discharge ("witch's' milk"). Neonatal breast hypertrophy usually resolves spontaneously within two weeks in boys and several months in girls. However, it may persist if the breast tissue is stimulated (eg, by attempting to express the milky discharge).Mastitis and/or breast abscess is another cause of breast enlargement in neonates. (17).

17.

1. Left parietal cephalhematoma, absorption of hematoma could lead to hyperbilirubinemia
2. meningomyelocele, encephalocele

Cephalohematoma is common in newborns and almost always resolves spontaneously with no any sequelae. Sometimes, complications such as hyperbilirubinemia, subdural hematoma, or infection due to skin laceration or erosion occur. Seldom there is intracranial hemorrhage. Infection of a cephalohematoma is rare, and the most common infecting organism was Escherichia coli (18).

18.

1. Contact dermatitis
2. Irritant contact dermatitis is a condition caused by direct injury of the skin. An irritant is any agent capable of producing cell damage in any individual if applied for

sufficient time and in sufficient concentration. Immunologic processes are not involved, and dermatitis occurs without prior sensitization. Irritants cause damage by breaking or removing the protective layers of the upper epidermis. They denature keratin, remove lipids, and alter the water-holding capacity of the skin. This leads to damage of the underlying living cells of the epidermis.

Irritant contact dermatitis consists of a spectrum of disease that ranges from a mild dryness, redness, or chapping to various types of eczematous dermatitis or an acute caustic burn. The severity of dermatitis produced by an irritant depends on the type of exposure, vehicle, and individual propensity. Normal, dry, or thick skin is more resistant to irritant effects than moist, macerated, or thin skin. Cumulative irritant dermatitis most commonly affects thin exposed skin, such as the backs of the hands, the web spaces of the fingers, or the face and eyelids. (19).

19.

1. As early as possible, the 1st day of life by neonatal screening
2.
   a. growth parameters
   b. developmental milestones

Immediate diagnosis and treatment of congenital hypothyroidism in the neonatal period is critical to normal brain development and physical growth. Treatment is usually effective if started within the first few weeks of

life. Delayed treatment may result in decreased intellectual capacity. Recommended treatment is lifetime daily administration of levo-thyroxine. Only the tablet form of levo-thyroxine should be prescribed. The U.S. Food and Drug Administration has not approved liquid suspensions. Suspensions prepared by pharmacists may lead to unreliable dosage. The tablets should be crushed daily, mixed with a few milliliters of water, formula or breast milk and fed to the infant. Levo-thyroxine should not be mixed with soy formula or with formula containing iron, as these products interfere with absorption ofthe medication. Dosage will need to be gradually increased as the infant grows. (20)

20.

1. Cerebral Palsy (CP) describes a group of conditions where movement and posture are affected as a result of damage to one or more areas of the brain. Cerebral palsy is characterized by an inability to fully control motor function, particularly muscle control and coordination. Types of cerebral palsy are as follows:

- Spastic (pyramidal): Increased muscle tone is the defining characteristic of this type. The muscles are stiff (spastic), and movements are jerky or awkward. This type is classified by which part of the body is affected: diplegia (both legs), hemiplegia (one side of the body), or quadriplegia (the entire body). This is the most common type of CP, accounting for about 70-80% of cases.

- Dyskinetic (extrapyramidal): This includes types that affect coordination of movements. There are 2 subtypes.
- Athetoid: The person has uncontrolled movements that are slow and writhing. The movements can affect any part of the body, including the face, mouth, and tongue. About 10-20% of cerebral palsy cases are of this type.
- Ataxic: This type affects balance and coordination. Depth perception is usually affected. If the person can walk, the gait is probably unsteady. He or she has difficulty with movements that are quick or require a great deal of control, such as writing. About 5-10% of cases of cerebral palsy are of this type.
- Mixed: This is a mixture of different types of cerebral palsy. A common combination is spastic and athetoid.

2. While specific therapies help a child develop specific skills and abilities, the overall goal of treatment is to help the individual with cerebral palsy reach his or her greatest potential physically, mentally, and socially. This is accomplished with a variety of different approaches managed by a team of professionals. Care for people with cerebral palsy is complicated, requiring a number of different services and specialists. In some areas, care is available through a single multidisciplinary clinic that oversees all aspects of the child's therapy.

a. Rehabilitation: physical therapy, special equipments, and spasticity treatment
b. Occupational therapy
c. Speech and language therapy
d. Ophthalmologist consultation

e. Treatment of seizures
f. Feeding and digestive problems
g. Breathing problems
h. Educational services
i. Medications for spasticity and abnormal movements: dopaminergic drugs, muscle relaxants (baclofen), benzodiazepines, botulinum toxin
j. Surgery: dorsal rhizotomy, implantation of a baclofen pump, stereotactic surgery, orthopedic surgical procedures. (21).

21. Polythelia (supernumerary nipple):

Supernumerary nipples (SNs) are a common minor congenital malformation that consists of nipples and/or related tissue in addition to the 2 nipples normally appearing on the chest. Supernumerary nipples are located along the embryonic milk line. Ectopic supernumerary nipples are found beyond the embryonic milk line. The embryonic milk line is the line of potentially appearing breast tissue as observed in many mammals. In humans, the embryonic milk line extends bilaterally from a point slightly beyond the axillae on the arms, down the chest and the abdomen toward the groin, and is generally thought to end at the proximal inner sides of the thighs, although supernumerary nipple has been described on the foot. Supernumerary nipples can appear complete with breast tissue and ducts and are then referred to as polymastia, or they can appear partially with either of the tissues involved.

Supernumerary nipple features are found in a number of syndromes, but, in most cases, it is probably a chance finding. These syndromes include Turner syndrome,

Fanconi anemia, and other hematologic disorders; ectodermal dysplasia; Kaufman-McKusick syndrome; and Char syndrome. Numerous sporadic publications linked supernumerary nipples to an association with anomalies or diseases, but such an association is probably only a chance finding.

Other associations of supernumerary nipples include the following:

- Central nervous system - Epilepsy, migraine, neurosis, familial alcoholism, fetal alcohol syndrome, intracranial aneurism, neural tube defect, developmental delay
- Gastrointestinal - Peptic ulcer, pyloric stenosis
- Ears, nose, throat and lung - Laryngeal web, ear abnormalities, accessory lung lobe
- Skeletal - Hand malformation, vertebral anomaly, absence of rib, coronal suture synostosis, hemihypertrophy, arthrogryposis, scalp defects and microcephaly
- Cardiac - Essential hypertension; conduction defect; bundle-branch block; patent ductus arteriosus; congenital heart disease, atrial septic defect, and ventricular septal defect

In the following decade, numerous publications supported the claim for a close association of supernumerary nipples and a renal anomaly, but many others could not find evidence to support such an association, which remains controversial. (22,23,24).

22. Peripheral radial anomalies:(25)

comprise of a large spectrum of anomalies which ranges from partial to a complete deficiency of the radius + / - bones of the thumb. They can be associated with a number

of associations which include:

- Rothmund-Thomson syndrome (RTS)
- TAR syndrome : thrombocytopaenia - absent radius
- Holt-Oram syndrome
- trisomy 18
- Fanconi anaemia
- Nager syndrome
- amniotic band syndrome
- VACTERL association
- Duane radial ray syndrome (DRRS)
- Cornelia de Lange syndrome (CdLS)
- omphalocoele-radial ray (ORR) complex
- in utero teratogen exposure
- valproic acid (valproate embryopathy)
- thalidomide (thalidomide embryopathy)

They are classified into four sub types depending on the extent of severity:

- type I : radius is slightly (> 2 mm) short and the hand to bends sideways at the wrist (often associated with a hypoplastic thumb) ; proximal radius usually unaffected
- type II : the radius bone is very short and the ulna curves sideways and supports the wrist poorly.
- type III : partial absence of radius
- type IV : complete absence of radius

23.

1. 12 months of age
2. The rationale for treatment of the undescended testicle is the prevention of potential sequelae. The most common

problems associated with undescended testicles are testicular neoplasm, subfertility, testicular torsion and inguinal hernia

Unlike the risk of testicular cancer, however, there seems to be an advantage to early orchiopexy for protection of fertility. Through testicular biopsy at the time of orchiopexy, germ cell density has been shown to decrease over time, beginning as early as one year of age. For this reason, treatment of the undescended testicle is recommended as early as six months of age and should be completed before age two. (26,27).

24.

The steps of the treatment of marasmus are:(28,29,30)

1. Nutritional management of the acute phase of severe marasmus (week 1)

This period corresponds to maintenance of vital functions and tissue renewal (ie, maintenance needs). During this period, the electrolyte imbalance, infections, hypoglycemia, and hypothermia are treated, and then feeding is started. Oral renutrition of a child with marasmus should be started as early as possible, as soon as the child is stable and the hydroelectrolyte imbalances are corrected. The term gut rest has no physiological basis. Enteral feeds decrease diarrhea and prevent bacteremia from bacterial translocation.

2. Rehabilitation phase (weeks 2-6)

In the rehabilitation phase of treatment, nutritional intake can reach 200 kcal/kg/d. The goal is to reach a continuous catch-up growth in weight and height in order to restore a healthy body weight. Only children who have been weaned

from their NG tube can be considered as being in the rehabilitation phase. Therefore, specific goals of this phase are as follows:

- To encourage the child to eat as much as possible
- To restart breastfeeding as soon as possible
- To stimulate the emotional and physical development
- To actively prepare the child and mother to return to home and prevent recurrence of malnutrition

25.

Right knee swelling and effusion:
a. trauma
b. septic arthritis
c. juvenile rheumatoid arthritis
d. Hemophilia

there is no treatment algorithm uniformly agreed upon for the management of JRA (Juvenile rheumatoid arthritis, Idiopathic rheumatoid arthritis). In general, NSAIDs can be used in all JRA types to control pain and diminish inflammation. Methotrexate at 15 mg/ m^{-2} /week should be instituted upon confirmation of the diagnosis of the polyarticular course of JRA. A short course of oral prednisone (0.5-2 mg kg^{-1}) may be required for particularly active forms of disease . If the response to methotrexate is inadequate, a biologic agent should be added to the drug regimen. However, almost no evidence exists on the various biologic agents, such as etanercept, adalimumab, abatacept, anakinra, infliximab, rituximab, and tocilizumab. Moreover, because of the lack of sound long-term safety data, the

existing evidence is insufficient to draw firm conclusions about the balance of the risks and benefits of using any biologic agent for treating JRA. Clinicians should be aware of this lack of evidence when considering biologics for patients with JRA.

Bone marrow transplantation has been initiated as an experimental treatment of severe autoimmune diseases including rheumatic diseases unresponsive to conventional therapy. Autologous stem cell transplantation (ASCT) is currently being evaluated, but only in a small number of children. This treatment approach is indicated only for children who are affected by severe active disease that fails to be controlled by conventional strategies, including anti-TNF therapy.(31)

26.

Wilson disease

1. Wilson's disease is diagnosed on the basis of the presence of at least two of the following criteria: 1. Family history of Wilson's disease; 2. KF rings; 3. low ceruloplasmin levels (<20mg/dL); 4. Free copper >25µg/dL [calculated as follows: free copper = serum copper (in mcg/dL) – (3 x ceruloplasmin in mg/dL)]; 5. 24-hour urine copper >100µg/24h; 6. copper in dry liver tissue.
   the copper in tissue assay can produce false-negatives in children, since it is dependent on sample size, the length of time during which the metal has been accumulating and the fact that it may be irregularly distributed.
2. The correct treatment should be initiated as early as possible in order to avoid or minimize the harmful effects of copper

accumulation in tissues. A diet restricting foods containing large concentrations of copper can help with treatment. Pharmacological treatment is with copper chelating drugs and the most widely used and studied is D-penicillamine, although it produces a series of side effects such as hypersensitivity, medullary depression, development of autoimmune diseases, neurological deterioration, nephrotoxicity, polyneuropathy, optic neuritis and polymyositis.(32,33,34,35).

27.

1. Right microphthalmia

Anophthalmia refers to complete absence of the globe in the presence of ocular adnexia (eyelids, conjunctiva, and lacrimal apparatus).

Microphthalmia refers to a globe with a total axial length (TAL) that is at least two standard deviations below the mean for age (see Table 1). For an adult eye, the lower 2.5% confidence limit for the TAL is about 21.0 mm. In a child in whom postnatal ocular growth continues into adolescence, the lower 2.5% confidence limit must be derived from a normative plot of TAL versus age .

Table 1. Length of the Neonatal and Adult Eye

	Posterior Segment Length	Anterior Segment Length	Total Axial Length
Neonate	10.2 mm	6.8 mm	17 mm
Adult	16.5 mm	7.3 mm	23.8 mm

1. Total axial length (TAL) is the axial distance (in mm) from the corneal apex to the back of the globe. 2. Anterior segment length (ASL) is the axial distance (in mm) from the cornea to the back of the lens. 3. Posterior segment length (PSL) is the axial distance (in mm) from the back of the lens to the back of the globe.

In microphthalmic eyes, measurements of ASL and PSL indicate that ASL is within or below the normal range, whereas PSL is uniformly at least two standard deviations below the mean for age . Most postnatal growth of the eye occurs in the first three years of life, particularly during the first year. Growth of the posterior segment accounts for 60% of the prenatal and more than 90% of the postnatal increase in TAL. Although TAL is reduced at birth, the microphthalmic eye can grow by a variable amount in the postnatal period depending upon the severity of the underlying malformation.

2.

a. Prenatal exposures associated with anophthalmia / microphthalmia include rubella, alcohol, thalidomide, retinoic acid
b. Trisomy 13
c. Mosaic trisomy 9
d. Microphthalmia with linear skin defects (MLS), also called MIDAS (microphthalmia, dermal aplasia and sclerocornea) and Gazali-Temple syndrome
e. Cerebro-oculo-facial-skeletal syndrome (COFS).(36)

28.

Cervical lymphadenopathy is a common problem in children. Lymphadenopathy posterior to the sternocleidomastoid is typically a more ominous finding, with a higher risk of serious underlying disease. In young children, anterior cervical lymph nodes as large as 2 cm, axillary nodes as large as 1 cm, and inguinal nodes as large as 1.5 cm in diameter are normal, and further evaluation is usually not indicated.(37,38,39).

A. Infectious etiologies:
1. viral infections: infectious mononucleosus, adenovirus, herpesvirus, coxsackie virus, cytomegalovirus, rubella, viral respiratory infections.
2. acute bacterial lymphadenitis: staph auerus, group A streptococcus
3. Atypical mycobacterial infections
4. streptococcal pharyngitis
5. mycobacterium tuberculosis

6. cat scratch disease(Bartonella henselae)
7. toxoplasmosis.

B. Non infectious etiologies:
1. malignant diseases: Hodgkin disease, non-Hodgkin lymphoma, leukemia, neuroblastoma, rhabdomyosarcoma
2. Kawasaki disease

The recommended investigations in cervical lymphadenopathy include:
1. CBC count and ESR
2. Skin testing for tuberculosis
3. Lymph node aspirate for culture
4. Titers for specific microorganisms: EBV, CMV, B hesnselae, toxoplasma, HIV
5. Chest radiography
6. Ultrasonography
7. Lymph node biopsy

29.

1. The embryological basis of neural tube defects:

Open NTDs have been suggested to result from defective primary neurulation while defective secondary neurulation gives rise to closed NTDs. However, this issue is not settled. Another possible explanation is that open NTDs (spina bifida in particular) result from defects in either primary or secondary neurulation, depending on their site being cranial or caudal to the posterior neuropore (ie, upper and lower spina bifida, respectively).

2.The possible underlying causes include:

a. genetic factors: A slight female predominance, and the higher incidence in certain ethnic groups and in the

offspring of consanguineous marriages, have suggested a genetic basis for NTDs. Chromosomal abnormalities (trisomy 13, 18, 21) also have been associated with NTDs. Concordance between monozygotic twins is low.

b. environmental factors: geographic location, season of conception, socioeconomic class, maternal diabetes, maternal age, zinc and folate deficiencies ,maternal alcohol abuse, maternal use of valproate, and intrauterine hyperthermia. carbamazepine monotherapy in the first trimester produces fetal malformations specific to spina bifida.(40,41,42,43,44).

30.

1.Serological markers of significance include EMA (antiendomysium IgA antibody)and tTG(anti-tissue transglutaminase IgA) antibodies. The sensitivity of tTG is 98% and specificity 96%, whereas the EMA is 100% specific and sensitivity is greater than 90%(45). Assays for tTG antibodies are largely based on the dominant antigen in the EMA test, however, tTG assays are more reliable and more reproducible, largely because the EMA is a qualitative assay and tTG assays are quantitative. The antibodies to tTG and deamidated gliadin peptide (DGP) have been combined in a multiplex immunoassay of persons suspected as having celiac disease, to potentially provide a complete antibody phenotype[46], and thereby to improve the performance characteristics of the serological testing. A meta-analysis has shown that the tTG antibody test out-performs the DGP antibody test, with a 5.2% greater sensitivity (93.0% *vs* 87.8%) and a 2.4% greater specificity (96.5% *vs* 94.1%), respectively[47].

2. Among the complications of undiagnosed and, therefore, untreated CD are growth failure in children, infertility, anemia, osteoporosis, small intestinal non-Hodgkin lymphoma[48], and a 3.9-fold increased all-cause mortality rate[49]. Potentially, this may underscore the importance of diagnosing and treating even latent CD.

Celiac patients were reported to have a 5.4-fold higher risk of non-Hodgkin's lymphoma, but no increased risk of Hodgkin's or chronic lymphatic leukemia. A shared susceptibility amongst siblings is observed. It remains controversial whether there is an increased risk of developing lymphoma in CD if the disease is asymptomatic[50].

There is a 5-fold increase in risk of lymphoproliferative malignancy in CD in comparison to the general population[51].

31.

- Digital clubbing
- Overlapping of toes

Overlapping toes are often familial, with the fifth toe being the most commonly affected. Frequently bilateral, the condition is distributed evenly between boys and girls. This deformity presents as adduction of the little toe with some external rotation of the digit. The metatarsophalangeal joint is dorsiflexed, and the nail plate is frequently smaller than expected.(52)

In newborns, the condition is frequently passively correctable with gentle stretching or use of various toe spacers. Nighttime paper-tape splinting to the adjacent toe

has been empirically recommended, but this method lacks evidence-based outcome studies.(53)However, if a child starts to walk before the deformity is corrected, it can become rigid, causing symptoms, and can require surgical correction.(54).

32.

1.

a. In 95% of cases of Down syndrome there are 3 copies of chromosome 21, the origin of the supernumerary chromosome is maternal in 97% of cases due to defect in meiosis I (non dysjunction)

b. Approximately 1% of individuals are mosaics with some cells having 46 chromosomes

c. Four percent of individuals have a translocation(the majority consist of fusion at the centromere between chm 13,14,15 or 21(Robertsonian translocation)

d. partial trisomy: very rare, a part of the long arm of chm 21 in triplicate

e. the least common, are Down syndrome patients without a visible chromosome abnormality

2.

a. hypothyroidism

b. diabetes mellitus(55,56).

33.

1. Hydrocephalus

a. large head **b.** sun-setting eyes **c.** right exotropia

2.

- A careful medical history
- A physical and neurological examination
- Cranial ultrasound
- Computerized tomography (CT)or magnetic resonance imaging of the brain (MRI) . (56,57,58).

34.

1. Holoprosencephaly (HPE)

2. Holoprosencephaly (HPE) occurs when an unborn baby's brain does not grow forward and divide properly during early pregnancy. Normally, the brain splits into two halves (hemispheres) during development. The hemispheres communicate to each other through a band of 200-250 million nerve fibers, called the corpus callosum. In patients with HPE, the hemispheres are not separated properly. As a result, there are abnormalities in the face and structure and function of the brain. Common signs and symptoms include a small head (microcephaly), excessive fluid in the brain (hydrocephalus), and intellectual disabilities (mental retardation) of varying degrees.

The severity of HPE ranges from mild to severe. According to studies, in about 97% of cases, the malformations are so severe that the baby dies before birth. In less-severe cases, babies are born with normal or near-normal brain development and facial malformations that may affect the nose, eyes, and upper lip.

According to the National Institute of Neurological Disorders and Stroke (NINDS), there are three classic subtypes of HPE: alobar HPE, semilobar HPE, and lobar

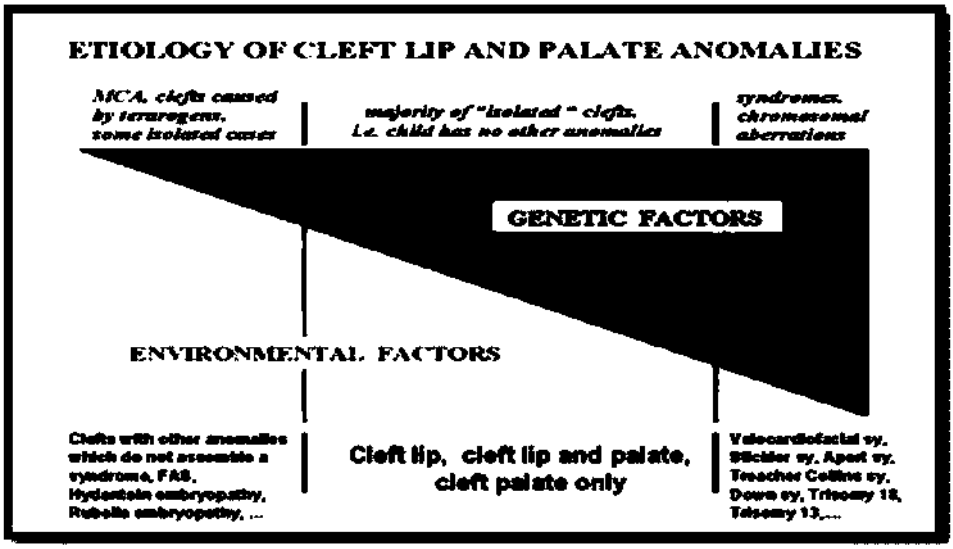
HPE. In alobar holoprosencephaly , there is a complete absence of midline forebrain division, resulting in a monoventricle and fused cerebral hemispheres. Semilobar holoprosencephaly is characterized by an incomplete forebrain division, resulting in partial separation of the cerebral hemispheres, typically posteriorly.

In lobar holoprosencephaly , there is complete ventricular separation, with focal areas of incomplete cortical division or anterior falcine hypoplasia present.(59,60,61).

35.

Bilateral cleft lip and palate

1. Cleft lip and cleft palate commonly occur as isolated birth defects, but are also associated with many genetic conditions.



2. most common treatment protocol presently used in most cleft treatment centers:

- Newborn - Diagnostic examination, general counseling of parents, feeding instructions, palatal obturator (if necessary); genetic evaluation and specification of diagnosis; empiric risk of recurrence of cleft calculated; recommendation of a protocol for the prevention of a cleft recurrence in the family
- Age 3 months - Repair of cleft lip (and placement of ventilation tubes)
- Age 6 months - Presurgical orthodontics, if necessary; first speech evaluation
- Age 9 months - Speech therapy begins
- Age 9-12 months - Repair of cleft palate (placement of ventilation tubes if not done at the time of cleft lip repair)
- Age 1-7 years - Orthodontic treatment
- Age 7-8 years - Alveolar bone graft
- Older than 8 years - Orthodontic treatment continues.(62,63)

36.

1. Large tongue (macroglossia)(64,65,66,67)
2. etiology:

a. Congenital causes

- Idiopathic muscle hypertrophy
- Gland hyperplasia
- Hemangioma
- Lymphangioma
- Down syndrome
- Beckwith-Wiedemann syndrome
- Behmel syndrome
- Lingual thyroid

- Gargoylism
- Transient neonatal diabetes mellitus
- Trisomy 22
- Laband syndrome
- Lethal dwarfism of Blomstrand
- Mucopolysaccharidoses
- Skeletal dysplasia of Urbach
- Tollner syndrome
- Autosomal dominant inheritance
- Microcephaly and hamartoma of Wiedemann
- Ganglioside storage disease type I

b. acquired causes

- Cretinism
- Diabetes
- Ludwig angina
- Acromegaly
- Neurofibromatosis
- Lingual thyroid
- Lymphangioma
- Hemangioma
- Carcinoma
- Plasmacytoma
- Amyloidosis
- Sarcoidosis

37.

1. Mongolian spots

More than 80% of black, Asian and east Indian infants have these lesion, whereas the incidence in white infants is less than 10%, they usually fade during the first few years of

life due to darkening of the overlying skin.

2. the peculiar hue of these macules is due to the dermal location of melanin-containing melanocytes that are presumably arrested in their migration from neural crest to epidermis. (68).

38.

1. The type of rash in measles is a red maculopapular eruption that begins around the forehead and spreads downward to the torso and extremities. The exanthema frequently becomes confluent on the face and upper trunk
2.
   a. pneumonia: giant cell pneumonia, superimposed bacterial infection, Bronchiolitis obliterans
   b. croup, tracheitis, bronchiolitis, otitis media
   c. diarrhea
   d. encephalitis
   e. subacute sclerosing parencephalitis(SSPE)

The risk factors for complications in measles include:

1. Age < 1year and >20 years
2. Crowding
3. Severe malnutrition
4. Low serum retinol levels
5. Immune compromise conditions(69).

39.

1. extremely unusual !!, the family of this child follow the instruction about rotation of sites of insulin injections.
2. In this condition , the mid-dermal collagen is replaced by hypertrophic fat cells on histopathologic sections .

Treatment of this condition involve frequent alteration of injection sites and the use of highly purified insulin.(70)

40.

Acrodermatitis enteropathica:

Acrodermatitis enteropathica (AE) is an inborn error of zinc metabolism that is inherited as an autosomal recessive disorder. Symptoms in infancy typically include periorificial and acral dermatitis , diarrhea, behavioral changes, and neurologic disturbances. In older children, failure to thrive, anorexia, alopecia, nail dystrophy, and repeated infections are most common.

Zinc deficiency may be due to inadequate intake, malabsorption, excessive loss, or a combination of these factors. The differential diagnosis include :

- Atopic Dermatitis
- Biotin Deficiency
- Candidiasis
- Epidermolysis Bullosa
- Protein-Energy Malnutrition
- In patients with acrodermatitis enteropathica (AE), zinc gluconate or sulfate is administered orally at a dosage of 1-3 mg/kg/d. Although the intravenous dosage has not clearly been estimated, amounts of 300-1000 mcg/kg/d may be sufficient for rapid reversal of symptoms. Clinical response is observed within 5-10 days. Warm compresses and petrolatum applied 3 times a day to areas of weeping or crusted dermatitis may enhance re-epithelialization when used concurrently with zinc replacement.(71).

41.

1. Preterm baby
2. Down syndrome
3. infant of diabetic mother
4. respiratory distress syndrome

The problems that infants of diabetic mothers are liable for are:

1. Hypoglycemia
2. Hypocalcemia and hypomagnesemia
3. Perinatal asphyxia
4. Polycythemia , hyperbilirubinemia, renal vein thrombosis
5. Repiratory distress syndrome
6. Cardiomegaly , heart failure, asymmetric septal hypertrophy
7. Macrosomia and birth trauma
8. Congenital anomalies: cardiac, lumbosacral agenesis, neural tube defects, renal, duodenal and anorectal atresia, situs inversus, holoprosencephaly, small left colon syndrome.(72).

42.

1. A collodian baby: a manifestation of congenital icthyosiform erythroderma or lamellar icthyosis.
2. Icthyosis is an inherited disorder of cornification, the morphologic abnormalities include hyperkeratosis, accumulation of lipid droplets within corneocytes and absence of normal lamellar granules. These granules have an important role in barrier formation and desquamation. Neonatal morbidity and deaths may result from:

a. Cutaneous infections
b. aspiration pneumonia
c. hypothermia
d. hypernatremic dehydration from excessive fluid losses. (73)

43.

1. Ranula(74,75,76)

Ranulas are mucoceles that occur in the floor of the mouth and usually involve the major salivary glands. Specifically, the ranula originates in the body of the sublingual gland, in the ducts of Rivini of the sublingual gland, in the Wharton duct of the submandibular gland, and, infrequently from the minor salivary glands at this location. Oral and plunging ranulas, if large, may affect swallowing, speech, or mastication and may result in airway obstruction. The very rare thoracic ranula may compromise respiratory function and may be life threatening.

2.The differential diagnosis include:

- Branchial Cleft Cyst
- Lingual thyroid, thyroglossal cyst
- Dermoid Cyst
- Lipomas
- Lymphangioma
- Oral Hemangiomas
- Oral Lymphangiomas
- Oral Pyogenic Granuloma
- Venous Lakes

44.

1. Gastroschisis

Gastroschisis may result from one of the following:

- Defective mesenchymal development at the junction of the body stalk and abdominal wall, resulting in increased abdominal pressure that may cause the dysplastic abdominal wall to rupture.
- Abnormal involution of the right umbilical vein or a vascular accident involving the omphalomesenteric artery may cause localized weakness and subsequent rupture.
- Rupture of a small omphalocele, absorption of the sac, and growth of skin between the resultant opening and the umbilical cord has been chronicled on prenatal ultrasonography.

2. babies with omphaloceles have associated abnormalities more frequently than gastroschisis, including intestinal problems (eg, Meckel diverticulum, intestinal atresia), genetic syndromes (eg, Beckwith-Wiedemann, trisomy 18), and congenital heart disease.(77,78,79,80)

45.

1.Cystic fibrosis

2. a Sweat test

- Several methods are used to conduct a sweat test. Performed properly, the quantitative pilocarpine iontophoresis test (QPIT) to collect sweat and perform a chemical analysis of its chloride content is currently considered to be the only adequately sensitive and specific type of sweat test. The sweat chloride reference value is less than 40 mmol/L, and a value of more than 60 mmol/L of chloride in the sweat is consistent with a diagnosis of cystic fibrosis. In babies aged 3 months or younger, results of 30-60 mEq/L are considered borderline and require retesting.

- Other causes of elevated levels of sweat chloride include the following:

- Untreated adrenal insufficiency
- Glycogen storage disease
- Type I fucosidosis
- Hypothyroidism
- Vasopressin-resistant diabetes insipidus
- Ectodermal dysplasia
- Malnutrition
- Mucopolysaccharidosis
- Panhypopituitarism
- Familial cholestasis
- Familial hypoparathyroidism
- Atopic dermatitis
- Iatrogenic causes (ie, infusion of prostaglandin E_1, improper technique)

- neonatal screening for cystic fibrosis. All screening algorithms in current use in the United States rely on testing for immunoreactive trypsinogen (IRT) as the primary screen for cystic fibrosis. The presence of high levels of IRT, a pancreatic protein typically elevated in infants with cystic fibrosis, warrants second level testing in the form of repeat IRT testing, DNA testing, or both.
- Chest radiography
- Genotyping
- More than 1600 cystic fibrosis mutations have been identified.
- A finding of 2 *CFTR* mutations in association with clinical symptoms is diagnostic, but negative results on genotype analysis do not exclude the diagnosis.
- Pulmonary function testing (PFT)

- Standard spirometry may not be reliable until patients are aged 5-6 years; however
- The recently described forced oscillation technique (FOT), which uses the impulse oscillometry system (IOS), can be used successfully in younger children.

*Bronchoalveolar lavage fluid usually shows a high percentage of neutrophils, and recovery of *Pseudomonas aeruginosa* from bronchoalveolar lavage fluid supports the diagnosis of cystic fibrosis in a clinically atypical case.

*Sputum microbiology: The most common bacterial pathogens in the sputum of patients with cystic fibrosis are *Haemophilus influenzae, Staphylococcus aureus, P aeruginosa, Burkholderia cepacia, Escherichia coli,* and *Klebsiella pneumoniae*. Findings of *P aeruginosa,* especially the mucoid form, support the diagnosis of cystic fibrosis in children.(81,82,83,84,85)

46.

1. **a.** Scoliosis
   b. Caffee au lait spot
2. Café au lait spots, or café au lait (CAL) macules (CALMs), are hyperpigmented lesions that may vary in color from light brown to dark brown; this is reflected by the name of the condition, which means "coffee with milk." The borders may be smooth or irregular. (86,87,88)

1. The most common associated systemic disorder is neurofibromatosis type 1 (NF1).
2. McCune-Albright syndrome: This syndrome often has one large, asymmetric café au lait macule with irregular borders, which is often described as being like the "coast

of Maine." The syndrome is associated with polyostotic fibrous dysplasia, which leads to pathologic fractures, precocious puberty, and numerous hyperfunctional endocrinopathies. Early in life, it may present with a single, large irregular café au lait spot. Follow-up observations reveal the endocrine abnormalities.

3. Fanconi anemia: Café au lait macules are present along with mental retardation, aplastic anemia, and risk for malignancy.
4. Tuberous sclerosis: Café au lait spots are present along with Ash leaf spots, facial angiofibromas, hemangiomas, cardiac rhabdomyomas, and shagreen patches.
5. Silver-Russell syndrome
6. Ataxia telangiectasia
7. Bloom syndrome
8. Basal cell nevus syndrome
9. Gaucher disease
10. Chiak-Higashi syndrome
11. Hunter syndrome
12. Maffucci syndrome
13. Multiple mucosal neuroma syndrome
14. Watson syndrome

47.

In recent years, IVIG has been used for numerous immunologically mediated conditions. In the presence of Rh, ABO, or other blood group incompatibilities that cause significant neonatal jaundice, IVIG has been shown to significantly reduce the need for exchange transfusions.

The 2004 AAP guidelines suggest a dose range for IVIG of 500-1000 mg/k .

The recommended dose is 500 mg/kg infused intravenously over a period of 2 hours for Rh or ABO incompatibility when the total serum bilirubin levels approach or surpass the exchange transfusions limits. On occasion, repeated the dose 2-3 times. In most cases, when this is combined with intensive phototherapy, avoiding exchange transfusion is possible. In the authors' institution, with about 750 NICU admissions per year, the use of exchange transfusions has decreased to0-2 per year following the implementation of IVIG therapy for Rh and ABO isoimmunization. The IVIG is not used in the presence of hydrops. Anecdotally, IVIG appears less likely to be successful when the infant is anemic (Hb < 10 g/dL).

Oral bilirubin oxidase can reduce serum bilirubin levels, presumably by reducing enterohepatic circulation; however, its use has not gained wide popularity. The same may be said for agar or charcoal feeds, which act by binding bilirubin in the gut. Bilirubin oxidase is not available as a drug, and for this reason, its use outside an approved research protocol probably is proscribed in many countries. (89,90)

48.

1. signs of respiratory distress
2. central cyanosis
3. single nostril

Craniofacial anomalies of holoprosencephaly include the following: close-set orbits (ocular hypotelorism) or infrequently, widely spaced ones (ocular hypertelorism); nasal abnormalities, ranging from flattened nose or deficient nasal septum to single nostril, choanal atresia, or arhinia;

thin lip with indistinct philtrum; cleft upper lip; cleft palate; small mouth; small mandible; or in rare cases, agnathia.(91).

49. Umbilical granuloma

Granulation tissue may persist at the base of the umbilicus after cord separation. The tissue is composed of fibroblasts and capillaries and can grow to more than 1 cm. Umbilical granulomas must be differentiated from umbilical polyps, which do not respond to silver nitrate cauterization. Small umbilical granulomas usually respond to silver nitrate application. Large umbilical granulomas or those that persist after silver nitrate treatment require surgical excision. (92,93)

50.

1. Left parotid abscess

   Acute parotitis in neonates

   This rare form of parotitis is lethal without treatment. In January 2004, Spiegel et al reviewed the literature and stated that only 32 cases had been reported in journals during the previous 3 decades. The characteristic clinical picture was of a sick premature infant with unilateral parotid swelling and inflammation. Seventy-five percent of the cases were in male infants. Pus expressed from the duct cultured *S aureus* in more than half of the cases. Most all of the cultured bacteria were from organisms present in the oral cavity, which suggests an ascending infection from the mouth.
2. Treatment is prompt administration of gentamicin and antistaphylococcal antibiotics plus adequate hydration, with a cure in approximately 80% of cases. Failure to

improve after 24-48 hours of treatment necessitates surgical drainage. Recurrence is uncommon.(94)

51.

1. Left facial palsy
2. Congenital facial paralysis is classified as traumatic or developmental, unilateral or bilateral, and complete or incomplete (paresis). Determining the etiology is important because the prognosis and treatment differ, depending on the underlying pathophysiology. More than 90% of patients with facial nerve paralysis caused by trauma recover without treatment. When the palsy is of developmental origin the parents should be informed that the child will never have an entirely normal appearance. The best outcome expected in these cases is facial symmetry at rest, near symmetry with voluntary movement, and spontaneous emotive movement. Much controversy exists regarding the timing of facial reanimation and the need for surgical exploration in children with congenital facial paralysis. Issues regarding the timing of reanimation are complex. Some health professionals advocate initial surgery during preschool to prevent the psychosocial aspects associated with a physical abnormality. However, waiting until adolescence when facial growth is mature and the child is able to understand the risks and benefits of surgery and participate in the decision making process also has merit.(95,96,97)

52.

1. Human herpes virus 6
2. Roseola is a common childhood disease. The causative organism is human herpesvirus 6 (HHV-6). The classic presentation of roseola infantum is a 9- to 12-month-old infant who acutely develops a high fever and often a febrile seizure. After 3 days, a rapid defervescence occurs, and a morbilliform rash appears. In roseola infantum, complications are rare. Given that seroconversion is practically universal, finding any of the complications that have been reported in the gastrointestinal, central nervous, pulmonary, and hematopoietic systems is rare. Children who have seizures with roseola are not expected to have further febrile or non febrile seizures.(98,99).

53.

1.Twin-to-twin transfusion

2. Twin-to-twin transfusion syndrome (TTTS) is the result of an intrauterine blood transfusion from one twin (donor) to another twin (recipient). TTTS only occurs in monozygotic (identical) twins with a monochorionic placenta. The donor twin is often smaller with a birth weight 20% less than the recipient's birth weight. The donor twin is often anemic and the recipient twin is often plethoric with hemoglobin differences greater than 5 g/dL. The donor twin becomes hypovolemic and oliguric or anuric. Oligohydramnios develops in the amniotic sac of the donor twin. Profound oligohydramnios can result in the stuck twin phenomenon in which the twin appears in a fixed position against the uterine wall. Ultrasonography typically fails to visualize the fetal bladder because of absent urine. The recipient

twin becomes hypervolemic and polyuric. Polyhydramnios develops in the amniotic sac of the recipient twin.

Either twin can develop hydrops fetalis. The donor twin can become hydropic because of anemia and high-output heart failure. The recipient twin can become hydropic because of hypervolemia. The recipient twin can also develop hypertension, hypertrophic cardiomegaly, disseminated intravascular coagulation, and hyperbilirubinemia after birth. Intrauterine demise of one twin can result in neurologic sequelae in the surviving twin. Acute exsanguination of the surviving twin into the relaxed circulation of the deceased twin can result in intrauterine CNS ischemia.

The most useful staging system for TTTS was developed by Quintero:

Stage	Oligohydramnios/ Polyhydramnios	Absent Urinein Donor Bladder	Abnormal Doppler Blood Flow	Hydrops Fetalis	Fetal Demise
I	+	-	-	-	-
II	+	+	-	-	-
III	+	+	+	-	-
IV	+	+	+	+	-
V	+	+	+	+	+

The most common procedure to treat TTTS is reduction amniocentesis. This procedure involves draining the amniotic fluid from around the recipient twin. This

procedure may improve circulation in the donor twin especially if the anastomosis are superficial in the placenta and the TTTS is a lower stage. This procedure may need to be performed multiple times during the pregnancy. Fetoscopic laser photocoagulation of chorionic plate vessels is a highly specialized procedure performed in a few centers around the world. This is mostly reserved for more severe cases, especially those that do not respond to amnioreduction.

Outcome is dependent upon gestational age at birth and whether intrauterine fetal brain ischemia occurred. The lower the gestational age at birth the greater the risk for long-standing neurologic or pulmonary sequelae. Catch-up growth occurs postnatally in most of the smaller donor twins. (100,101,102).

54.

1. Right Erbs palsy
2. Many cases of BPP(Brachial plexus palsies) are transient, with the child recovering full function in the first week of life. A smaller percentage of children continue to have weakness leading to long-term disability from the injury. The mainstay of treatment for these children is physical and/or occupational therapy in concert with a regular home exercise program. A select few patients may benefit from surgical intervention in the early stages to improve innervation of the affected muscles. Others benefit from tendon transfers performed later to improve shoulder and (sometimes) elbow function.

Numerous other nonsurgical treatments, including electrical stimulation and botulinum toxin injections, also may prove effective in the treatment of children with BPP.

Statistics on children who attain complete recovery after brachial plexus palsy (BPP) vary widely, with reports ranging from 4-93%. This discrepancy is due, at least in part, to the time of evaluation. Many children present to the newborn nursery with temporary weakness (neurapraxia) that resolves prior to discharge and is thus unaccounted for in most of these studies. Peripheral nerves remyelinate at a rate of 1 mm/day. Thus, if the nerve is not transected, recovery can be expected by 4-5 months in Erb's palsy, 6-7 months for an upper-middle trunk palsy, and 14 months for a total BPP. Some authors believe that infants who do not show signs of spontaneous recovery by 3-5 months usually are left with residual deficits if managed conservatively. As one might expect, findings consistent with a more extensive initial injury (Horner's syndrome and total BPP) portend a less favorable prognosis. The converse also is true; children with isolated upper trunk lesions generally have a better prognosis. The presence or absence of phrenic nerve involvement does not appear to have prognostic value in BPP.

Yilmaz and coworkers compared MRI, electrophysiologic studies, and muscle strength scoring in 13 infants with BPP to determine which indicator provided the most accurate prognosis of the outcome at 1 year. They found that scoring of muscle strength (eg, elbow flexion; wrist, finger, and thumb extension) was the most reliable measure, with 94.8% confidence at 3 months. Electrophysiologic studies, while helpful in identifying patients with an unfavorable prognosis, occasionally are overoptimistic (in 1 of 13 cases). MRI findings of pseudomeningoceles were seen in 2 of 5 patients with an unfavorable prognosis and in 2 of 8 with a

good prognosis.(103,104,105).

55.

1. Blue sclera
2. Blue sclera is characterized by localized or generalized blue coloration of sclera because of thinness and loss of water content, which allow underlying dark choroid to be seen.

1. Associated with high urine excretion
   - **a**. Folling syndrome (phenylketonuria)
   - **b**. Hypophosphatasia (phosphoethanolaminuria)
   - **c.** Lowe syndrome (oculocerebrorenal syndrome;chondroitin-4-sulfate-uria)
2. Associated with skeletal disorders
   - **a**. Brachmann-de Lange syndrome.
   - **b.** Brittle cornea syndrome (blue sclera syndrome)-recessive
   - **c.** Crouzon disease (craniofacial dysostosis)
   - **d.** Hallermann-Streiff syndrome (dyscephaliamandibulooculofacial syndrome)
   - **e.** Marfan syndrome (dystrophia mesodermalis congenita)
   - **f.** Marshall-Smith syndrome
   - **g**. McCune-Albright syndrome (fibrosus dysplasia)
   - **h.** Mucopolysaccharidosis VI (Maroteaux-Lamy syndrome)
   - **i.** Osteogenesis imperfecta (van der Hoeve syndrome)
   - **j** Paget syndrome (osteitis deformans)
   - **k.** Pierre Robin syndrome (micrognathia-glossoptosis syndrome)
   - **l.** Robert syndrome
   - **m.** Silver-Russell syndrome
   - **n.** Werner syndrome (progeria of adults)

3. Chromosome disorders
   a. Trisomy syndrome
   b. Turner syndrome
4. Ocular
   a. Congenital glaucoma
   b. Myopia
   c. Repeated surgeries
   d. Scleromalacia (perforans)
   e. Staphyloma
   f. Trauma
5. Miscellaneous
   a. Ehlers-Danlos syndrome (fibrodysplasia elastic generalisata)
   b. Goltz syndrome (focal dermal hypoplasia syndrome)
   c. Incontinentia pigmenti (Bloch-Sulzberger syndrome)
   d. Lax ligament syndrome
   e. Minocycline-induced
   f. Oculodermal melanocytosis (nevus of Ota)
   g. Pseudoxanthoma elasticum (Grönblad-Strandberg syndrome)
   h. Relapsing polychondritis (106,107,108)

56.

1. Ambigous genitalia
- .3-Beta-Hydroxysteroid Dehydrogenase Deficiency
- 5-Alpha-Reductase Deficiency
- Androgen Insensitivity Syndrome
- Congenital Adrenal Hyperplasia
- Denys-Drash Syndrome
- Genital Anomalies

- Gonadoblastoma
- Hypogonadism
- Hypopituitarism
- Hypospadias
- Microphallus
- Precocious Puberty

2.Chromosomal analysis

a. Endocrine screening

b. Serum chemistries/electrolyte tests

c. Androgen-receptor levels

d. 5-alpha reductase type II levels

e. Renal/bladder ultrasonography

f. Genitography

g. h. CT scanning and MRI are usually not indicated but may help identify internal anatomy.

i. Exploratory laparotomy/gonadal biopsy

j. Diagnostic laparoscopy/gonadal biopsy: Histologic analysis of gonadal biopsy specimens may identify ovarian tissue, testicular tissue, ovotestes, or streak gonads.(109,110)

57.

1. Anemia in an adolescent girl
   - **a.** Iron deficiency anemia
   - **b.** Anemia of chronic disease
   - **c.**Hemoglobinopahies
2. The recommended investigations are:
   - **a.** complete blood counts, red blood cells morphology, reticulocyte count
   - **b**. Serum Iron, total iron binding capacity and serum

ferritin

c. hemoglobin electrophoresis and measurement of hemoglobin F and A2

d. Testing stool for the presence of hemoglobin

e. Reticulocyte hemoglobin content (CHr): measurement of CHr may be a reliable method to assess deficiencies in tissue iron supply and, in combination with a CBC, may be an alternative to the traditional biochemical panel for the diagnosis of iron deficiency in children(111)

f. Bone marrow aspiration.

58.

1. **a.** premature baby
   b. very low birth weight
   c. signs of respiratory distress (subcostal recession)
   d. facial and legs bruising and echymosis
2. **a.** respiratory distress syndrome
   b. apnea of prematurity
   c. hypoglycemia, hypocalcemia, hyperbilirubinemia
   d. intraventricular hemorrhage
   e. necrotizing enterocolitis
   f. sepsis.

59.

Post term baby: long nails, meconium stained nails and umbilical Cord.

60.

Rickets (rachitic rosary):

Treatment for rickets may be administered gradually over several months or in a single-day dose of 15,000 mcg

(600,000 U) of vitamin D. If the gradual method is chosen, 125-250 mcg (5000-10,000 U) is given daily for 2-3 months until healing is well established and the alkaline phosphatase concentration is approaching the reference range. Because this method requires daily treatment, success depends on compliance.

If the vitamin D dose is administered in a single day, it is usually divided into 4 or 6 oral doses. An intramuscular injection is also available. Vitamin D (cholecalciferol) is well stored in the body and is gradually released over many weeks. Because both calcitriol and calcidiol have short half-lives, these agents are unsuitable for treatment, and they bypass the natural physiologic controls of vitamin D synthesis.

The single-day therapy avoids problems with compliance and may be helpful in differentiating nutritional rickets from familial hypophosphatemia rickets (FHR). In nutritional rickets, the phosphorus level rises in 96 hours and radiographic healing is visible in 6-7 days. Neither happens with FHR.

If severe deformities have occurred, orthopedic correction may be required after healing.

61.

1. Achondroplasia
2. Operation for pyloric stenosis

Achondroplasia, hypochondroplasia, and metaphyseal chondrodysplasias are considered short-limb dwarfing conditions. These patients' sitting height is within normal range. Additional terms used to describe the segment of the

limb with the greatest involvement are rhizomelic (proximal), mesomelic (middle), and acromelic (distal). In achondroplasia, the extremity involvement is rhizomelic, with the arms and thighs more severely involved than the forearms, legs, hands, and feet.(113)

The primary defect found in patients with achondroplasia is abnormal endochondral ossification. Periosteal and intramembranous ossification is normal. Tubular bones are short and broad, reflecting normal periosteal growth. The iliac crest apophyses (appositional growth) are normal, giving rise to large, square iliac wings. The growth of the triradiate cartilage (endochondral growth) is abnormal, giving rise to horizontal acetabular roofs. Thus, these patterns of defect help to explain many of the observed clinical and radiographic characteristics of achondroplasia.

62.

Viral exanthem(macular and maculopapular exanthem

1. Measles
2. Rubella
3. Erythema infectiosum
4. Roseola infantum
5. Epstein-Bar virus and Aminopenicillins

63.

Cyanotic congenital heart disease

1. transposition of great arteries
2. tricuspid atresia
3. total anomalous pulmonary venous return
4. tetralogy of Fallot.

64.

Abdominal mass:

1. non Hogkins lymphoma
2. teratoma
3. neuroblastoma
4. mesenteric tumors and cysts.

65.

Harrison sulcus (groove):

1. Rickets
2. severe chronic asthma

66.

Multiple vertebral and rib anomalies with kyphoscoliosis

Hemivertebra is a rare congenital abnormality of the spine where only onenly one side of the vertebral body develops, leading to deformation of the spine,such as such as scoliosis or kyphosis. It results from failure of a vertebra to form on one side,ne side, resulting in a laterally based wedge vertebra with half a vertebral body,a single a single pedicle, and hemilamina. The cause of hemivertebra is unknown. The distribution pattern pattern of the anomaly does not implicate a specific environmental or genetic factor factor. Hemivertebra should be differentiated from the other vertebral abnormalities malities (wedge vertebra, butterfly vertebra, bloc vertebra, bar vertebra or any or any combination) that cause congenital scoliosis.

67.

Asymmetry of skin folds on the inner aspects of the thighs : suggestive of DDH

(developmental dysplasia of the hips), Ultrasonography (US) is the preferred modality for evaluating the hip in infants who are 6 months or younger. US enables direct imaging of the cartilaginous portions of the hip that cannot be seen on plain radiographs .Furthermore, US enables dynamic study of the hip with stress maneuvering. Plain radiographs of the pelvis are most helpful when significant ossification of the capital femoral epiphyses has occurred and when adequate US evaluation cannot be performed. Plain radiographs of the pelvis are obtained in the frontal projection with the legs in the neutral position. (115)

68.

Depressed skull fracture. It is advisable to elevate severe depressions to prevent cortical injury from sustained pressure.

69.

Mucopolysaccharidosis

Dysostosis multiplex refers to a constellation of skeletal abnormalities in MPS conditions diagnosed based on plain radiographs. Dysostosis multiplex is classic in Hurler syndrome . These findings include the following:

- Large skull with thickened calvaria, premature suture closure, j-shaped sella turcica, and shallow orbits
- Abnormal spacing of teeth with dentigerous cysts
- Short, thickened and irregular clavicles
- Short, wide, and trapezoid shaped phalanges

- Oar-shaped ribs
- Anterior hypoplasia of the lumbar vertebrae with kyphosis
- Poorly formed pelvis with small femoral heads and coxa valga
- Enlarged diaphyses of long bones and irregular metaphyses
- vertebral body beaking (in Morquio syndrome).

70.

Facial bruising, subconjunctival hemorrhage, Erythema, abrasions, echymosis of facial or scalp tissues may be seen after forceps or vacuum assisted deliveries.

71.

Severe neonatal anemia

Anemia in the newborn results from three processes

– Loss of RBCs: hemorrhagic anemia

a. Loss of placental integrity : abruption, previa

b. Anomalies of the umbilical cord or placental vessels

c. Twin-to-twin transfusion syndrome

d. Fetomaternal hemorrhage

e. Traumatic delivery (visceral, intracranial hemorrhage)

f. Defect in the hemostasis: congenital, thrombocytopenia, Vitamin K deficiency, iatrogenic blood loss.

– Increased destruction: hemolytic anemia:

a. Immune hemolysis: RH, ABO, autoimmune

b. Non immune: sepsis, TORCH infections

c. Congenital erythrocyte defects: G6PD,spherocytosis,elliptocytosis, unstable hemoglobin, thalassemia

d. Systemic disease: galactossemia, vitamin E deficiency

– Underproduction of RBCs: hypoplastic anemia:

a. Congenital: Diamond-Blackfan, congenital leukemia, sideroblastic anemia

b. Acquired: infections, aplastic anemia.

72.

The new lines of treatment in Thalassemia are:

1. Hematopoietic stem cell transplantation (HSCT)
2. Investigational agents known to increase Hb F level: This therapeutic strategy is investigational at this time. Several agents administered to raise the Hb F level have been investigated in patients with severe thalassemia. Unfortunately, the initial results of these studies are not promising. However, in one study, hydroxyurea was given to 11 patients with transfusion-dependent Hg E/β thalassemia. Treatment was effective in 4 patients who became transfusion independent; 4 other patients required less transfusions, and 3 patients did not respond.
3. Gene therapy: This therapy is an attractive therapeutic modality, the efficacy of which remains to be demonstrated. (116).

73.

Bilateral club foot

The true etiology of congenital clubfoot is unknown, most infants who have clubfoot have no identifiable genetic, syndromal, or extrinsic cause.

Extrinsic associations include teratogenic agents (eg, sodium aminopterin), oligohydramnios, and congenital constriction rings. Genetic associations include mendelian inheritance (eg, diastrophic dwarfism; autosomal recessive pattern of clubfoot inheritance).

Cytogenetic abnormalities (eg, congenital talipes equinovarus [CTEV]) can be seen in syndromes involving chromosomal deletion. It has been proposed that idiopathic CTEV in otherwise healthy infants is the result of a multifactorial system of inheritance. Evidence for this is as follows:

- Incidence in the general population is 1 per 1000 live births.
- Incidence in first-degree relations is approximately 2%.
- Incidence in second-degree relations is approximately 0.6%.
- If one monozygotic twin has a CTEV, the second twin has only a 32% chance of having a CTEV. (117,118).

74.

Pitting edema in a neonate

1. prematurity
2. erythroblastosis fetalis
3. congenital nephrotic syndrome
4. Hurler syndrome.

75.

1. Right facial palsy
2. Central cyanosis

patients with tetralogy of Fallot face additional risks that include paradoxical emboli leading to stroke, pulmonary embolus, and subacute bacterial endocarditis. It is well known that children with congenital heart disease are prone to stroke. In most of these children the causes of stroke have been related to thromboemboli, prolonged hypotension/anoxia and polycythemia. What is often forgotten is that residual shunts or a patent foramen ovale are also known causes of strokes. The investigation of strokes in these children usually begins with a CT scan of the brain followed by an ECHO. (119).

76.

Recent discoveries of the molecular basis of epidermolysis bullosa have resulted in the development of new diagnostic tools, including prenatal and preimplantation testing. Based on a better understanding of the basement membrane zone (BMZ) and the genes responsible for its components, new treatments (eg, gene or protein therapy) may provide solutions to the skin fragility found in patients with epidermolysis bullosa.

Epidermolysis bullosa is classified into 3 major categories, including (1) epidermolysis bullosa simplex (intraepidermal skin separation), (2) junctional epidermolysis bullosa (skin separation in lamina lucida or central BMZ), and (3) dystrophic epidermolysis bullosa (sublamina densa BMZ separation, as in the images below).

Researchers have proposed a new category termed

hemidesmosomal epidermolysis bullosa, which produces blistering at the hemidesmosomal level in the most superior aspect of the BMZ. Epidermolysis bullosa simplex usually is associated with little or no extracutaneous involvement, while the more severe hemidesmosomal, junctional, and dystrophic forms of epidermolysis bullosa may produce significant multiorgan system involvement.

Significant progress has been achieved in finding specific molecular therapies for epidermolysis bullosa, including protein and gene therapy. Type VII collagen and laminin-5 gene therapy have been proven effective through in vivo models. Type VII collagen protein therapy has similarly been shown to be effective in an in vivo model. Currently, these therapies are being extensively studied at the preclinical stage, in animal models (120).

Research therapies

Potential future therapies include protein and gene therapies. Model systems using these approaches show promise for significant advances in future therapies.

In protein therapy, the missing or defective protein is produced in vitro by recombinant methods and applied directly to blistered skin. Protein therapy may be most useful in epidermolysis bullosa subtypes involving a defect or deficiency in type VII collagen because this protein appears to have a long half life in the body.

In gene therapy, the goal is to deliver genes targeted to restore normal protein production. Gene therapy for one patient with a nonlethal form of junctional epidermolysis bullosa has been successful at the 1-year mark. This was

accomplished using a retroviral gene transfer system, using ex vivo gene transfer and grafting corrected keratinocytes back onto the patient.

Molecular therapy

Gene therapy for non-lethal junctional epidermolysis bullosa has been performed and shown to be efficacious in a small trial of one patient. In this trial, cultured patient keratinocytes received a normal copy of the LAMB3 gene through retroviral delivery, then the corrected cells were grafted back to areas of patient skin. Analysis over one year showed continued high expression of laminin 5 and a clinical absence of blistering.

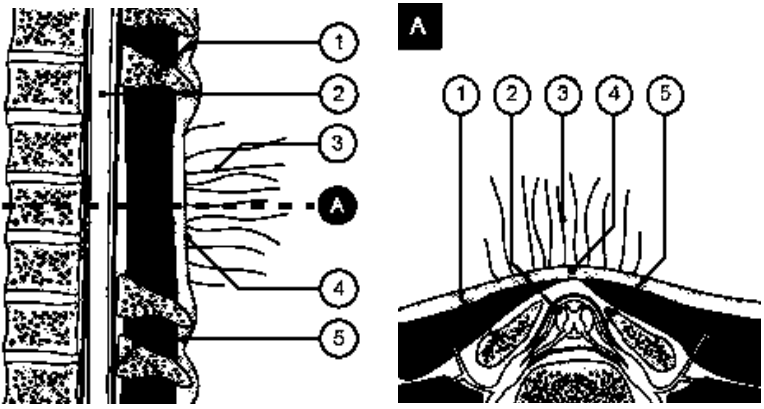
Two clinical trials for treatment of recessive dystrophic epidermolysis bullosa are currently underway. In one center at the University of Minnesota, bone marrow transplantation is used as the mechanism of delivery of corrective skin cells. In this trial, recessively inherited epidermolysis bullosa patients undergo bone marrow ablation and immunosuppression. The other clinical trial, which is being performed at Stanford University, consists of retroviral mediated type VII collagen gene transfer. In this trial patient skin cells are treated with type VII collagen gene in a retrovirus, and the cells are grafted back to the patient. Both studies are currently in progress and no peer reviewed scientific data are currently available (121).

77.

1. Spina bifida occulta with a tuft of hair at the site of the defect

2.Spina bifida occulta occurs very frequently and is usually found accidentally in x- rays or in an examination of the back. They seldom have clinical relevance because only a missing closure of the osseous structures exists in the formation of the vertebral arch, without the spinal cord with its membranes (meninges) being involved. The skin covering is intact. Sometimes a tuft of hair tells where the osseous structures are missing.

(longitudinal section) (cross section at the level of A)



1-Spinous process	**1-Vertebral arch**
2-Spinal cord	**2-Spinal cord**
3-Tuft of hair	**3-Tuft of hair**
4-Skin	**4-Skin**
5-Dura mater	**5-Dura mater**

Schematic drawing of a spina bifida occulta

The meninges and the spinal cord are in their right places. Optically, only a tuft of hair gives its presence away. since there is a problem with cell differentiation/migration that the skin over the area with the incomplete vertebra develops hair like that on the scalp since the appropriate hair growth inhibitory factors are not present.

78.

1.Prune Belly syndrome(122)

Prune belly syndrome is a group of birth defects that involve three main problems:

- Poor development of the abdominal muscles, causing the skin of the belly area to wrinkle like a prune
- Undescended testicles (cryptorchidism)
- Urinary tract problems

In 1839, Frölich first described prune belly syndrome (PBS), and Osler gave the condition its name. Prune belly syndrome is also called Eagle-Barrett syndrome and triad syndrome.

Children with prune belly syndrome can present with myriad renal, ureteral, and urethral abnormalities. Obstruction and/or upper urinary tract dilatation is not unusual in these children. The site of obstruction can vary from as high as the pelviureteral junction to as low as the prostatic membranous urethra.

Urinary tract abnormalities include

- bilateral hydroureteronephrosis : often with extremely dilated, tortuous ureters
- varying degrees of renal dysplasia
- enlarged urinary bladder, often with urachal diverticulum

- vesicoureteral reflux is common
- poor bladder contractility
- dilated posterior urethra without urethral obstruction.

2. Associations(123)

- aneuploidic syndromic associations
- trisomy 18
- trisomy 13
- other associations
- congenital cardiac anomalies
- ventricular septal defect (VSD)
- atrial septal defect (ASD)
- tetralogy of Fallot
- intestinal malrotation
- imperforate anus
- polydactyly / syndactyly
- talipes equinovarus

A lack of abdominal muscles leads to a poor cough mechanism, which, in turn, leads to increased pulmonary secretions. Weak abdominal muscles lead to constipation because of an inability to perform the Valsalva maneuver, which helps push the stool out of the rectum during defecation.

The mortality rate associated with prune belly syndrome is 20%. Prune-belly syndrome occurs with variable degrees of severity. In severe cases, renal dysplasia and oligohydramnios in utero result in pulmonary hypoplasia. These infants may be stillborn or die shortly after birth often due to respiratory complications.

Those with less severe renal disease may survive infancy, but may have recurrent urinary tract infection or progressive renal insufficiency. Some mild cases may have little or no

loss of renal function and therefore a better prognosis.
N.B this child had also mucopolysaccharidosis.

79.

1. Pierre _Rubin Syndrome: (124,125).

Robin sequence (RS), previously known as Pierre Robin syndrome and Pierre Robin anomalad, consists of the following 3 essential components:

- Micrognathia or retrognathia
- Cleft palate (usually U-shaped, but V-shape also possible)
- Glossoptosis, often accompanied by airway obstruction: The tongue is not actually larger than normal, but because of the small mandible, the tongue is large for the airway and therefore causes obstruction. Rarely, the tongue is smaller than normal.

2. The primary concern in airway compromise is its life-threatening aspect. Most neonates have an isolated defect that is not part of a syndrome, for which the airway and feeding complications are usually greater. The great majority of neonates can be treated in the prone position (face down). Devices or procedures such as oral airways, palatal prostheses, continuous positive airway pressure or endotracheal intubation, mechanical ventilation, and tracheostomy can be avoided. In patients who consistently maintain CO2levels above 50, a surgical procedure is appropriate. Three surgical procedures are used to treat Robin sequence: tongue-lip adhesion/glossopexy, tracheotomy, and distraction osteogenesis of the mandible. the tongue-lip adhesion/glossopexy has been a relatively controversial

technique. Nevertheless, it does have its isolated advocates among maxillofacial surgeons. Distraction osteogenesis is a relatively new technique for treating airway obstruction in Robin sequence. Distraction osteogenesis has been popularized by Jim Sidman, who has the world's widest experience in treating patients with Pierre Robin sequence with distraction osteogenesis.

Many centers have developed expertise in this area. In this technique, the mandible is cut near the angle of the mandible on both sides. A mechanical device distracts the 2 portions of the mandible approximately 1.5-2 mm a day. As the portions of the mandible are separated, new bone is formed, and the mandible gradually elongates over a period of 2-3 weeks. Distraction can be performed in the newborn to prevent a tracheotomyor can be performed later to remove a tracheotomy tube. Because this new technique has been popularized only during the last 5-10 years, the long-term sequelae on mandibular growth and tooth development is not known at this time; nevertheless, it remains a very promising technique that has been gaining in popularity (see the images below).

Distraction osteogenesis is a relatively new technique for treating airway obstruction in Robin sequence. Distraction osteogenesis has been popularized by Jim Sidman, who has the world's widest experience in treating patients with Pierre Robin sequence with distraction osteogenesis.

80.

1. von Willebrand disease (VWD) (126)
2. Screening tests for von Willebrand disease (VWD) include the following:

- CBC count: Assess platelet number and morphology, which should be normal in most patients with von Willebrand disease, except those with type 2B von Willebrand disease.
- Template bleeding time: Although neither sensitive nor specific for von Willebrand disease, template-bleeding time is outside of the reference range in about 50% of patients with type 1 von Willebrand disease. Patients with von Willebrand disease types 2A, 2B, 2M, and 3 often have prolonged bleeding times. The template bleeding time has largely been replaced by automatic platelet function analyzers (PFAs) such as the PFA-100.
- Prothrombin time (PT) is within reference range in von Willebrand disease.

- Activated partial thromboplastin time (aPTT): Approximately 25% of patients with type 1 von Willebrand disease have aPTT results outside of the reference range. Because aPTT and the template bleeding time are insensitive tests for von Willebrand disease , add von Willebrand factor (VWF) activity to the screening tests performed for patients with suspected bleeding disorders
- von Willebrand factor levels: von Willebrand factor levels vary and can be influenced by numerous factors including blood type. Individuals with type O blood have lower values of von Willebrand factor levels on average, whereas those with type AB blood have higher values of von Willebrand factor. Day-to-day variation in von Willebrand factor levels is a normal occurrence in the same individual; therefore, a single level within reference range does not exclude the diagnosis of von Willebrand disease.
- FVIII activity: FVIII activity is variably decreased.
- von Willebrand factor activity (ristocetin cofactor
- von Willebrand factor antigen
- platelet von Willebrand factor analysis . Gene analysis can also be performed for diagnosis.

81.

Microtia

Severe malformations of the external ear are rare, but minor deformities are common. Isolated abnormalities of the external ear occur in approximately 1% of children. The term microtia may indicate subtle abnormalities of the size,

shape, and location of the pinna and ear canal, or major abnormalities with only small nubbins of skin and cartilage and absence of the ear canal opening; Microtic ears often are more anterior and inferior in placement than normal auricles, and the location and function of the facial nerve may be abnormal. Cosmetic reconstruction of the auricle usually is performed between 5-7 yr of age, and is performed before canal atresia repair in children deemed appropriate for this surgery.(127).

82.

1- Congenital Arhinia

2-The treatment of arhinia after birth typically involves treatment of airway obstruction and feeding difficulties. This aspect of management is of extreme importance to pediatric surgeons. Since neonates are obligate nasal breathers, simultaneous sucking and breathing requirement in neonates with arhinia leads to respiratory distress. Inspiration and expiration through the oral passage alone may result in thoracic retraction, thereby further exacerbating respiratory distress. Temporary measures like oral airway and orogastric feeding are successful. Canalization of nasal passage or tracheostomy may be required depending on severity of neonatal respiratory distress(127,128,130).

Surgical reconstruction is very demanding and needs a team of pediatric, neuro, craniofacial, and plastic surgeons.

The components of repair include creation of orifice and reconstruction of external nose. Anterior aperture can be created by external approach and posterior orifice may be formed with endoscopic guidance(128,129).

Reconstruction of external nose should be done near adolescence because growth of the reconstructed nose is unpredictable if done earlier. But the psychological trauma to the parents and the child may demand early correction which can be done earliest by the age of 4–5 years. It includes reconstruction of bony and cartilage framework along with mucosal lining and skin coverage of external nose(127,130). Cartilage framework from costal cartilage placement in forehead and transfer of forehead flap with grafted framework for nose reconstruction is the most preferred option. Vertical distraction osteogenesis represents a modality for elongation of the mid face(128,131).

Treatment of craniosynostosis and hypertelorism is done around 1-year of age before the brain growth is hampered. The procedures involved are various types of advancement cranioplasty(128).

Antenatal ultrasound, preferably three dimensional with facial profile, can detect the condition earliest by 12–16 weeks of gestational age. Prenatal diagnosis has been reported in only one case during the second trimester of pregnancy(132). Prognosis of such children is poor in terms of mental and physical development as well as cosmetic and functional outcomes. Antenatal diagnosis at an appropriate

gestational age can provide the option of termination of pregnancy to the parents, especially if associated with other anomalies and trisomy.

One case has been reported in which simultaneous reconstruction of both the internal and external nose was undertaken in the newborn period(133).

Congenital arhinia is a rare anomaly and management is difficult. This case highlights the complexity of the condition with respect to surgical management. A multidisciplinary approach is essential along with expert neonatal medical and surgical care for the management.

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