

Class 3

1. An infant presented with a history of high-pitched, mewing cry, closely resembling the cry of a kitten, which lasted several weeks, and then disappeared. The affected child is hypotonic, has low birth weight, microcephaly, a round face with wide-set eyes, downward slanting of the palpebral fissures, strabismus, and a broad-based nose. The ears are low-set, abnormally shaped, and have narrow external auditory canals and preauricular tags.

What is the diagnosis?

2. 20-year-old patient presents with broad nasal bridge, flat face, oblique palpebral fissure, wide spaced eyes that look almond shape, large protruding tongue, small, low set ears, simian crease (single palmar crease), short neck, short stature wide space between 1 and 2 toes.

What is the main pathology in this patient?

3. Infant presents with weak cry, muscle hypotonia and marked hypoplasia of skeletal muscles and subcutaneous fat. There is poor sucking and low birth weight. Also, present is microcephaly, triangular face, hypoplastic orbital ridges, prominent nasal bridge, short palpebral fissures, microstomia, microtia, malformed ears, microstomia and micrognathia, retrognathia, short stature, clenched fingers, broad chest without wide spaced nipples
- Which of the following is the most likely condition?

4. Small for gestational age infant with holoprosencephaly (failure of the forebrain to divide properly), cleft lip and cleft palate, microphthalmia, colobomas and retinal dysplasia. Low set ears, hearing loss. Single transverse palmar crease, polydactyly, dextrocardia with cardiac anomalies. Cryptorchidism or bicornuate uterus. What is the most likely diagnosis?

5. A 20-year-old male patient is in your office for infertility treatment. The patient is tall with disproportionally long extremities, small and firm testes, gynecomastia. There is decreased spermatogenesis, decreased testosterone production, and increased estradiol in blood. There is no deep voice, no beard, no male hair distribution.
Which one of the following conditions is most likely present in this patient?
6. A 20-year-old female presents with undeveloped breasts, little pubic hair, short stature, webbed neck, shield like chest with wide spaced nipples and lymphedema of extremities
Which is the most likely condition in this patient?
7. A 4-year-old child that was normal at birth presents with intellectual disability and hyperactivity. The patient also has light sensitivity and incidence of multiple pyogenic infection every year. Patient examination shows light skin and hair, eczema, increased incidence of keratosis pilaris, musty or mousy odor.
What is the most likely condition?
8. A 6-month-old patient who was developing normally presents with developing mental retardation, decreased vision, motor and sensory deterioration and cherry red spots in the macula
What is the most likely cause?
9. A 6-month-old patient who was developing normally presents with developing mental retardation, decreased vision, motor and sensory deterioration and cherry red spots in the macula, and hepatomegaly.
What is the diagnosis?
10. An 18-month-old female with previous normal development is presenting with impaired language, problems walking, decreased ability to hold object, hand wringing, tapping (disappears when child sleeps). Loss of language
Repetitive head movement, toe walking, wide based stiff walk
What is the most likely disease?

11. The 17-year-old male patient presents with autistic like behavior. The patient has a long and narrow face, large and protruding ears, prominent chin and forehead, high arched palate and flexible fingers. The patient also has macrorchidism, poor speech, poor eye contact

What is the most likely disease?

12. A 50-year-old male presents with depression, apathy, irritability, and anhedonia. These symptoms started one year ago. At this time patient also is complaining of jerky hyperkinetic, dystonic movements in all parts of the body. On examination, the patient has bizarre, puppet-like gait and facial grimacing. Which one of the following is the most likely cause?

13. A 6-year-old patient presents with cutaneous and subcutaneous round painful masses, seven medium-brown, freckle-like macule, Lisch nodules, scoliosis, erosive bone defects, cystic bone lesions, and GI bleeding
Most likely cause is

14. A 18-year-old male patient presents with complaints of severe muscle stiffness, dysphagia and constipation. Examinations show sustained involuntary contractions, abnormal gait, foot dorsiflexion weakness, weakness of hand muscles, atrophy of facial muscle, ptosis and gonadal atrophy.

Which one of the following is the most likely cause?

15. A 5-year-old boy presents with complaints of palpitations, and weakness. During examination, the patient is pale with yellow sclera, percussion reveals splenomegaly. Blood work shows increased bilirubin, iron, MCHC and ferritin.

Which is the most likely cause?

16. A 50-year-old male patient presents with easy, recurrent, and excessive bruising, frequent nose bleeds, prolonged bleeding from minor skin trauma. The physical examination shows petechiae and knee hematomas. Patient also has noticed blood in the urine.

Which of the following is the most likely condition?

17. A 50-year-old tall male patient presents with long extremities, long fingers and toes, lax joints, and ligaments. There is also dolichocephalic head (long head), prominent supraorbital ridges and bilateral subluxation of the lens. Which is the most likely cause?

18. A 16-year-old male patient presents with frequent shoulder joint dislocations, falls and balance issues. The patient also has hyperextensible skin, loose ligaments, hypermobile joints. During examination skin is white and soft with a velvety feel, and underlying vessels are visible. The skin has a doughy feel. The skin is easily hyperextensible. It is easy to pull, and, once released, it immediately returns to its original state. The patient bruises easily.

The most likely cause?

19. A 10-year-old boy presents with multiple bone fractures, blue sclera, joint laxity, and scoliosis.

What is the most likely diagnosis?

20. A 20-year-old patient presents with Xanthelasma palpebrarum, Achilles tendon xanthoma and xanthomas on metacarpophalangeal extensor tendons. Patient has highly elevated LDL in blood.

The most likely cause is:

21. A 12-year-old patient presents with multiple chronic infections in respiratory tract, lung hyperinflation, atelectasis, airway obstruction – cough, wheezing, shortness of breath, sputum production, nasal polyps, malabsorption, intestinal obstruction, pancreatitis, pancreatic insufficiency, Cirrhosis, metabolic acidosis.

What is the most likely condition?

22. A 45-year-old male presents with slowly progressive dyspnea, cough, sputum production, wheezing. Patient history reveals childhood hepatitis that resolved. Patient does not smoke.

What is the most likely condition?

23. A 35-year-old male patient presents with jaundice, asymmetrical tremor, Kayser-Fleischer rings, difficulty speaking, excessive salivation, ataxia, masklike facies, clumsiness with the hands, and personality changes.

What is the most likely condition?

24. A 20-year-old female patient presents with complaints of fatigue, lethargy, and joint pain. During examination, there is diffuse skin hyperpigmentation and koilonychia

What is the most likely condition?

25. A patient presents with gait ataxia, followed by hand clumsiness, dysarthria, depressed deep tendon reflex, impaired vibration sensation, joint position, arrhythmias, congestive heart failure Gait ataxia – progressive slow and clumsy walking, after normal walking has developed. Difficulty standing, running Speech becomes slurred, slow, eventually incomprehensible

What is the most likely disease?

26. Male Infant presents with small tonsils and absence of lymph nodes, with recurrent pyogenic infections in lungs, sinuses, skin with Streptococcus pneumoniae, Hemophilus influenzae Pyogenic (pus) infections – lung, skin, CNS, joints

What is the most likely cause?

27. Male that presents with: Bleeding into tissues – muscle hematomas, hemarthrosis, retroperitoneal hemorrhage/ Trivial hit to the head – intracranial hemorrhage Pain (produced by beginning of bleeding) platelet, PTT, and PT, Coagulation factor assay – determines diagnosis A of B, von Willebrand factor

What is the most likely disease?

28. A previously healthy male patient presents with sudden onset of abdominal pain and fatigue. Patient started to take primaquine as a prophylaxis before

upcoming trip 3 days earlier. During examination there is scleral icterus and paleness. Blood work shows low hemoglobin and elevated bilirubin. Blood smear shows bite cells and Heinz bodies.

What is the most likely disease?

29. A 35-year-old male patient that presents with c/o vision problems, decreased urination and edema. The patient's history shows hearing loss in childhood and development of hematuria at age of 25. During examination, there is noticeable nystagmus.

What is the most likely disease?

30. A child presents with developmental delay and issue with the knees. The patient's knees swell and are painful. Knee symptoms get better with allopurinol. The child also has episodes of self-harming, like cutting his hands. The examination reveals poor muscle tone and involuntary upper extremity movement.

Which of the following is the most likely condition?

31. A 5-year-old boy presents behavioral problems at school and home. Physical examination shows protruding tongue, gargoylism, and hepatosplenomegaly. Patient's hearing is also decreased. Blood work shows elevated heparan and dermatan sulfate

What is the condition?

32. A patient presents with complaints of tiredness, fatigue, muscle pains, sore tongue and dizziness. The patient also has diarrhea and palpitations. The patient also states that urine has become dark. During examination, T is 101 F, P 110 b/min, RR 18, BP 100/70 mm/Hg. Blood shows elevated bilirubin.

What is the most likely disease?

33. Patient presents with nausea, vomiting, fullness after eating, abdominal pain. Lately patient also feels tired and weak. Endoscopy shows low gastric acidity and blood work shows large red blood cells.

What is the most likely disease?

34. Patient presents with easy bruising, bleeding from nose, gums, development of petechia, blood in urine, splenomegaly, increased bleeding time.

What is the most likely disease?

35. A male patient presents with hematuria, edema, high blood pressure, bloody sputum., cough, dyspnea, malaise, weight loss, fatigue, fever, and chills. Blood work shows uremia.

What is the most likely disease?

36. A 40-year-old female patient presents with fluctuating muscle weakness. Patient states her strength improves after rest but starts to decline with activity and is much worse at the end of the day. The patient states that it is particularly hard to look at the computer screen.

What is the most likely disease?

37. A 40-year-old female presents with fatigue, general weakness. She always feels hot and has increased thirst, hunger and frequency of urination. During examination, her skin is moist, warm to touch. HR is 120, BP 150/90.

What is the most likely disease?

38. Patient presents with increased sleepiness, weight gain, loss of appetite, constipation, hoarseness of voice and feeling cold. Slow movement peripheral neuropathy Sleepiness, slow speech, day time somnolence Sleep apnea Menstrual irregularities Macroglossia Nonpitting edema of arms and legs Bradycardia

What is the most likely disease?

39. A 40-year-old female patient presents with recurrent miscarriages, red cheeks connecting over bridge of the nose. Maculopapular rash over exposed areas of skin, Fever, weight loss, fatigue. Intermittent joint pains (arthralgias) proteinuria (glomerulitis to membranoproliferative glomerulonephritis), edema, hypertension, abdominal pain

What is the most likely disease?

40. A 40-year-old female presents with fever, malaise, multiple joint pains, during examination there is symmetrical hand joint swelling and pain on palpation.

What is the most likely disease?

41. A 30-year-old female presents with dry mouth, dry eyes, parotid gland. Increased risk of developing lymphoid malignancies. Arthritis, arthralgia, myalgia, anemia, leukopenia, neuropathy, vasculitis, renal tubular acidosis, dry skin.

What is the most likely disease?

42. Patient presents with fever, malaise, night sweats, loss of appetite, weight loss. Patient also complains of arthralgia, myalgia, neuralgia, altered sensation, headaches, seizures, strokes, oliguria, hypertension, uremia, hematuria, proteinuria, abdominal pain, (stomach, liver, GB, pancreas ischemia and necrosis) - nausea, vomiting, bloody diarrhea, skin ulcer, tender erythematous nodules.

What is the most likely disease?

43. Female patient presents with very tight skin, dramatically decreased mobility of digits, mask-like face and cannot move lips. Polyarthritis – fingers, wrists, dysphagia.

What is the most likely disease?

44. A 6-month-old baby presents with systemic candidiasis infection, recurrent diarrheas, viral infections and failure to thrive.

What is the most likely disease?

45. Male infants that present in the first year of life with severe bloody diarrhea and persistent thrombocytopenia. Patient has frequent nose bleeds, mucous bleeds and eczema. At this moment patient also has purulent cough and nasal discharge with fever of 102 F. Labs show low T

cell count, Low IgM levels, high IgE and IgA, normal IgG, and neutrophil malfunction.

What is the most likely disease?

46. A 5-year-old child presents with history of recurrent infections since birth. During examination, patient presents with long face, high and broad nasal bridge, midline facial cleft, a small receding mandible, small teeth. Patient also has behavioral and communication problems. What is the most likely disease?