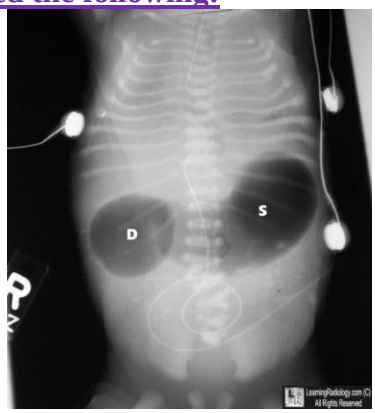
- **Case (1): one day infant with bilious vomiting.**
  - **What is you differential diagnosis?**
    - ✓ Duodenal atresia.
    - ✓ Jejunal atresia.
    - ✓ Ileal atresia.
    - ✓ Anal atresia.

  On physical examination the neonate appears to be jaundiced, has tachycardia (with grade-II systolic murmur), no cyanosis, abdomen is not distended, and there is generalized hypotonia (there is no head-lag reflex).
  - **What are the three features of intestinal obstruction?**
    - ✓ Abdominal distention. This patient has no distended abdomen which indicates that the obstruction is proximal (in duodenum).
    - ✓ Vomiting.
    - ✓ Constipation.

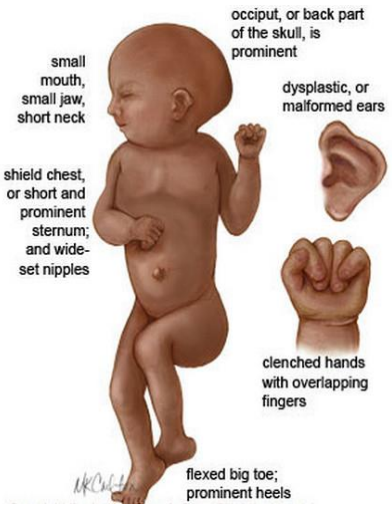
  X-ray was done and it showed the following:



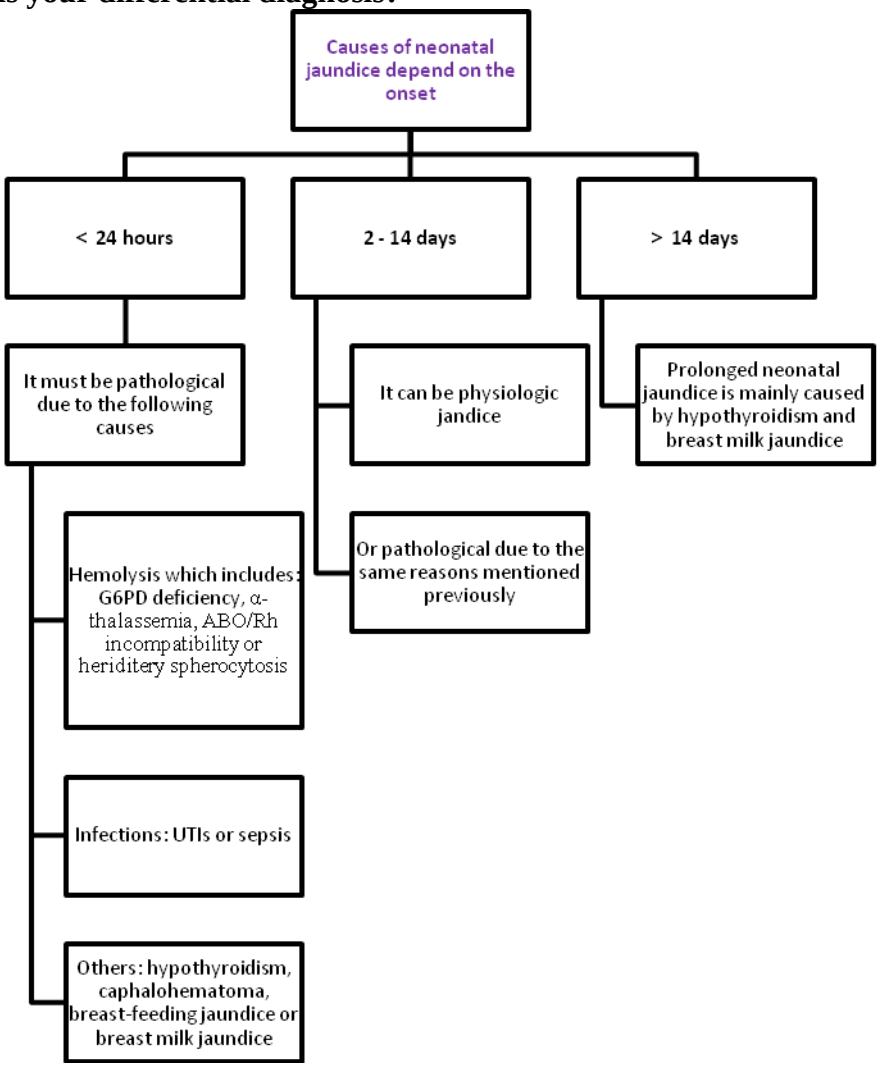
  - **This is called double-bubble sign. Therefore, your definitive diagnosis will be duodenal atresia (which can be associated with Down's syndrome).**
- **Case (2): an infant presents with the following on inspection**

  - **The images show (from left to right):** cleft palate, polydactyli, scalp defect.
  - **What is you diagnosis**? Patau syndrome (trisomy 13).
  - **How would you confirm your diagnosis?** karyotyping.
- **Case (3): What are the features of Edward's syndrome (trisomy 18)?**
  - Mental retardation and hypertonia.
  - Small facial features.
  - Clenched hands with overlapping digits.
  - Rocker bottom feet.

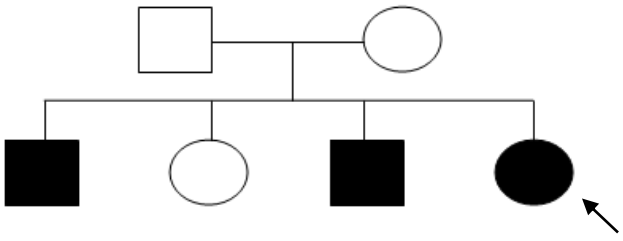
    Notice that 95% of cases die within the first year of life.



- **Case (4): 3 days female with jaundice since 2 days. The mother had two previous babies with history of jaundice.**
  - **What is your differential diagnosis?**

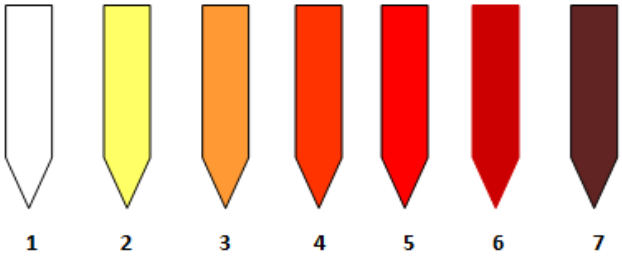


    - ✓ <u>As the mother had 2 previous babies with the same condition we have to look for a recurrent cause:</u>
      - ❖ It can be due to α-thalassemia.
      - ❖ ABO (not possible).
      - ❖ Rh incompatibility (negative Coomb's test).
      - ❖ UTI, hypothyroidism and infections are all non-recurrent causes.
  - **The definitive diagnosis is**: ↓G6PD activity.



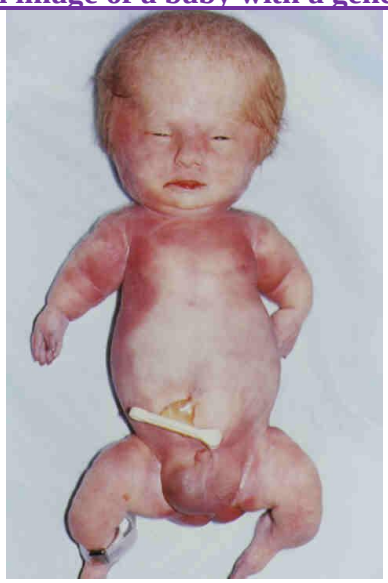
    - ✓ <u>Remember that the mode of inheritance of G6PD deficiency is X-linked recessive → then how could a female (our case) be affected with the disease?</u>
      - ❖ Either both parents are affected.
      - ❖ Or the mother is a carrier and the father is affected (but have never experienced any symptoms).
      - ❖ Or due to Turner's syndrome (monosomy X).
      - ❖ Or due to skewed X-inactivation.

- **Case (5): a 2 years old boy with the following urine samples.**



- **Describe what you see.**
  - ✓ There are seven samples of urine placed in order; sample (7) shows red color urine which is decreasing in color until being clear as seen in sample (1).
- **What is your diagnosis?**
  - ✓ Hemoglobinuria due to hemolysis caused by G6PD.
- **What is your treatment?**
  - ✓ Blood transfusion.

- **Case (6): The following is an image of a baby with a genetic disease:**



- **X-ray was done and it showed**: severe osteopenia with multiple fractures in long bones and ribs.
- **What is your diagnosis?**
  - ✓ Osteogenesis imperfect: an autosomal dominant disease (usually inherited due to a new mutation) and characterized by abnormal type-I collagen. Clinical features are:
    - ❖ Blue sclera.
    - ❖ Fragile bones (causing knock-knees and osteoporosis).
    - ❖ Yellow or gray-blue teeth.
    - ❖ Easy brusability.

- **Case (7): see the image and mention your diagnosis.**

- **This is achondroplasia**, an autosomal dominant disease which is mainly characterized by rhizomelia (proximal long bone abnormalities: humerus and femur). Clinical features include: frontal bossing, mid-face hypoplasia, lumbar lordosis and trident-shaped hands.