Introduction:
More than 50 cases of this lethal disorder have been reported since the initial Description by Albers-Schonberg\(^1\) in 1907. It is a rare metabolic disease that is characterized by diffuse increase in skeletal density and obliteration of marrow spaces. It is one of variety of skeletal dysplasia (predominantly metaphyseal and diaphyseal changes). The skeleton shows cores of calcified cartilage that are surrounded by areas of new bone, this new bone formation is normal but there is a deficiency of bone and cartilage resoption. The osteoclasts are abnormal and have been noted to lack functional ruffled border.\(^3\)

Two types are distinguished:
1. Congenital (Juvenile, malignant, or infantile) which is of autosomal recessive form\(^2\) and characterized by severe anemia, hepatosplenomegaly, pancytopenia, cranial and optic nerve palsy and a compromised immune system, hence, repeated haemorrhage and infection usually leads to death in early childhood.\(^4\)
2. Adult (Tarda) form. It is inherited as an autosomal dominant trait \(^5\) but some investigators report that autosomal recessive forms of this disease exist as well. Although osteopetrosis tarda is much less severe than the infantile form, a Lifelong history of fractures usually characterized the clinical picture. The patients are also prone to bone infection, cranial nerve palsy due to bone encroachment on foramina, also may found.

Case report:
A 3 months old male infant is the last child in a family of 8 children, 5 females and 3 males all are phenotypically normal delivered from consanguineous parents (father 45 years and mother 38 years old) with history of 2 females deathes, one died at the age of 2 months, cause of death unknown. The other one at the age of 4 years, cause of death: osteopetrosis.
- Preconceptional and prenatal history: no history of exposure to mutagens or teratogens.
- Obstetric history: FTND-
- Neonatal history: no history of neonatal jaundice, cyanosis or RD.

Physical examination:
Head circumference, high and weight are normal. Has abnormal facial features, mild hypertelorism, broad nasal bridge, abnormal shape of the ear, abnormal eyes, R1 facial palsy, high arched palate and postaxial polydactyly, chest clinical clear, heart NAD and hepatosplenomegaly.
Investigation:
Skeletal survey: The bones, including the base of the skull, ribs, pelvis and metaphyseal ends of long bones show increased density.

References: