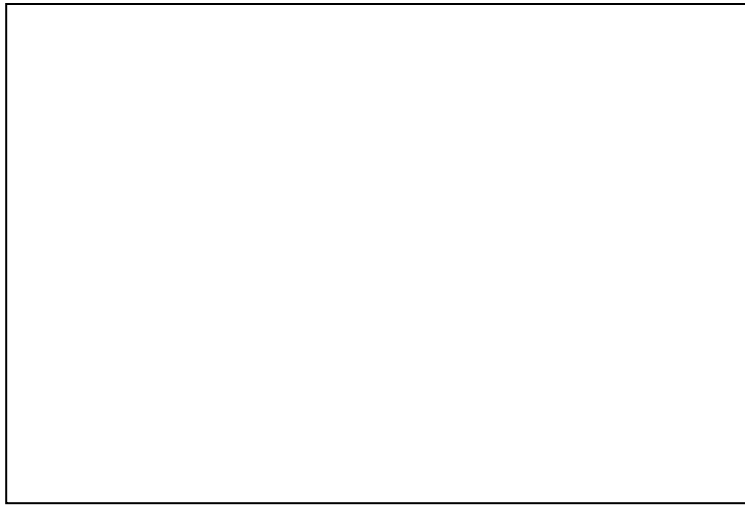


MEDICAL QUIZ

Sheikha Al-Arrayed, PhD

Seven months old male baby from non-consanguineous family was seen in the genetic clinic. He had short stature, macrocephaly, prominent forehead, low nasal bridge, rhizomelic form of short limb, stubby trident hands, lordotic lumbar spine, very short lower limbs, inversion of both feet, short and malposition of fibula, undescended testis, Prominent buttocks and hypotonia. The mother is 26 year old with juvenile diabetes.



- Q1. What is your diagnosis?
- Q2. What are the investigations?
- Q3. What is the differential diagnosis?
- Q4. What are the main complications of this condition?
- Q5. What is the inheritance?