



Web Fig. 1: **A:** Clinical photograph of proband's uncle showing the deep set eyes, prominent zygomatic arches, low set ears, swollen bilateral knee joints and anteriorly bowed tibiae. **B:** Radiograph of the pelvis of the proband's uncle showing osteopenia and protrusio acetabulae (marked by red arrow). **C:** Radiograph of the left knee of the proband's uncle suggestive of osteopenia. **D:** Clinical photograph of proband's aunt showing microcephaly, deep set eyes, corneal scarring of left eye, prominent zygomatic arches and low set ears. **E:** Radiograph of the pelvis of proband's aunt showing osteopenia. **F:** Radiograph of the thoracolumbar spine of proband's aunt showing relatively milder degree of osteopenia and exaggerated thoracic curve. **G:** Clinical photograph of proband's sister showing microcephaly, deep set eyes, prominent zygomatic arches, bony prominence over the left lateral supra orbital margin and low set ears. **H:** Radiograph of the pelvis of proband's sister showing osteopenia. **I:** Radiograph of the thoracolumbar spine of proband's sister showing a relatively milder degree of vertebral compression. **J:** Clinical photograph of proband showing microcephaly, deep set eyes, asymmetric palpebral fissures, squint, prominent zygomatic arches and low set ears. **K:** Radiograph of the pelvis of proband showing marked osteopenia. **L:** Radiograph of the thoracolumbar spine, lateral view of proband showing osteopenia and vertebral compression (marked by arrows).