

Fig. 1 Clinical photograph and radiography images of patient 2. **(a)** Small palpebral fissures bilaterally. **(b)** 3D surface-rendered MR images demonstrate small palpebral fissures and relatively underdeveloped tarsal plate of left lower eyelid (open arrow). **(c)**, **(d)** T2W axial MR images at the level of orbits show normal globes and optic nerves.

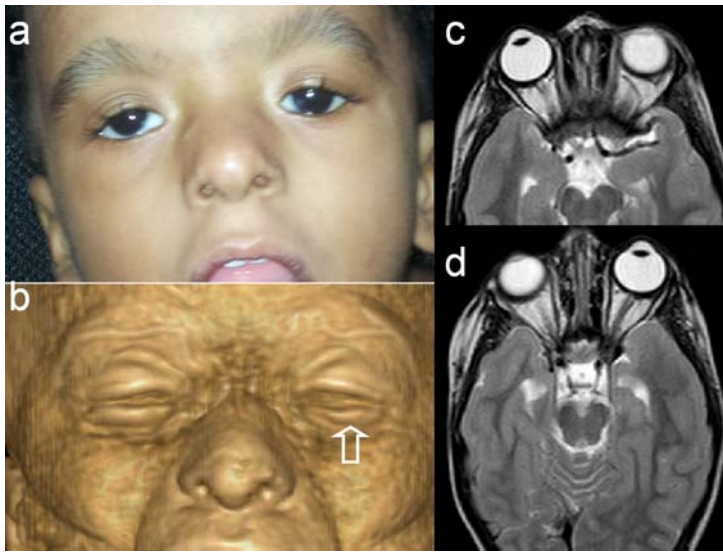


Fig. 2 Clinical photograph and radiography images of patient 2. (a) Prominent winged right scapula. (b) Chest radiograph does not show structural abnormality of scapulae. (c) 3D MR image viewed from behind shows high right scapular spine (open arrow).

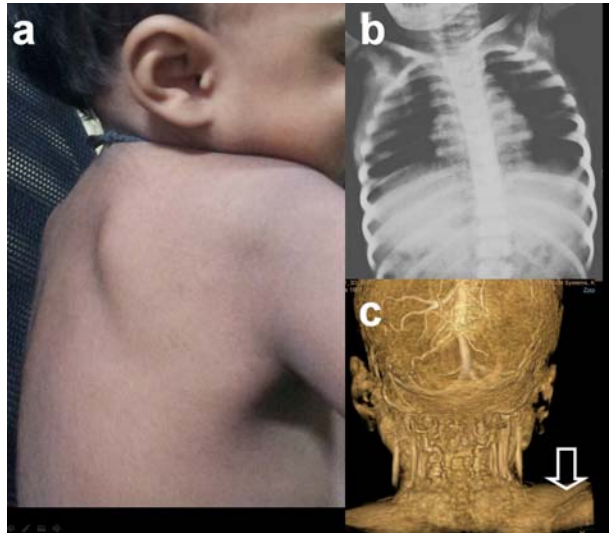


Fig. 3 Integrative genomics viewer snapshot showing hemizygous variant, c.887G>A in exon 7 of *MED12* in the proband (<http://software.broadinstitute.org/software/igv>).

