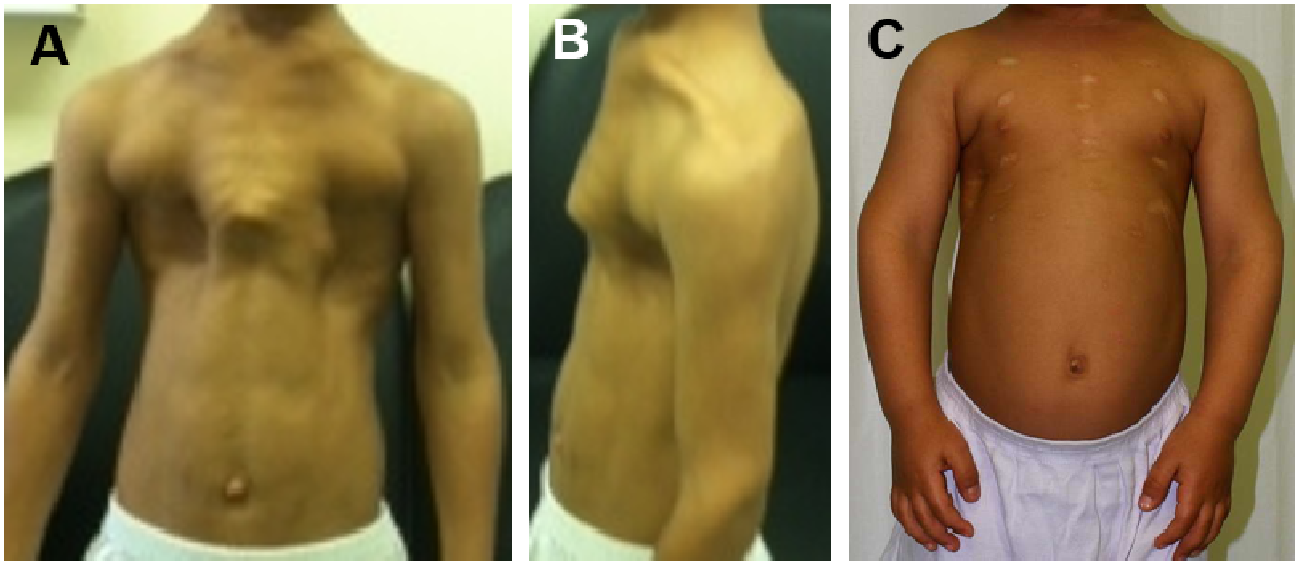

S1 Fig. Clinical features of axial spondylometaphyseal dysplasia (axial SMD) patients with *C21orf2* mutations.

(A), (B) P7, 9-year-old boy. Very narrow chest, severe Harrison sulci with pectus carinatum. (C) P1-2, 5-year-old boy. Mild chest deformity. Both are homozygotes of the same *C21orf2* mutation, c.643-23A>T.