Supplementary material to article by C. E. Zimmermann et al. "Schöpf-Schulz-Passarge Syndrome: Previously Unreported WNT10A Genotype and Phenotypes in 9 Family Members”

Fig. S2. Graphical illustrations of the WNT10A mutations detected.
The WNT10A gene comprises four exons and is located on the long (q) arm of chromosome 2 (2q35). It encodes a protein of 417 amino acids belonging to a large family of secreted signalling proteins. (A) Wild-type protein sequence of WNT10A (417 aa). (B) Sequence map of exon 2 showing the c.321C>A; p.Cys107* mutation in the human WNT10A gene. (C) Sequence map of exon 3 showing the c.742C>T; p.Arg248* mutation in the human WNT10A gene. Both nonsense mutations introduce premature stop codons TGA (indicated by a box).