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

A. Wildtype WNT10A (417 aa)

MGSAHPRPWLRLRPQPQPRPALWVLLFFLLLLAAAMPRSAPHDILDLRLPPEPVLNANTV CLTLPGLSRRQMEVCVRHPDVAASAIQGIQIAIHECQHQFRDQRWNCSSLETRNKIPYES PIFSRGFRESAFAYAIAAAGVVHAVSNACALGKLKACGCDASRRGDEEAFRRKLHRLQLD ALQRGKGLSHGVPEHPALPTASPGLQDSWEWGGCSPDMGFGERFSKDFLDSREPHRDIHA RMRLHNNRVGRQAVMENMRRKCKCHGTSGSCQLKTCWQVTPEFRTVGALLRSRFHRATLI RPHNRNGGQLEPGPAGAPSPAPGAPGPRRRASPADLVYFEKSPDFCEREPRLDSAGTVGR LCNKSSAGSDGCGSNCCGRGHNILRQTRSERCHCRFHWCCFVVCEECRITEWVSVCK*

в. WNT10A exon 2 (c.321C>A;p.C107*) G G ٨ G MGSAHPRPWLRLRPQPQPRPALWVLLFFLLLLAAAMPRSAPNDILDLRLPPEPVLNANTV CLTLPGLSRRQMEVCVRHPDVAASAIQGIQIAIHECQHQFRDQRWN* C. WNT10A exon 3 (c.742C>T;p.R248*) G ٨ MGSAHPRPWLRLRPQPQPRPALWVLLFFLLLLAAAMPRSAPNDILDLRLPPEPVLNANTV CLTLPGLSRRQMEVCVRHPDVAASAIQGIQIAIHECQHQFRDQRWNCSSLETRNKIPYES PIFSRGFRESAFAYAIAAAGVVHAVSNACALGKLKACGCDASRRGDEEAFRRKLHRLQLD ALQRGKGLSHGVPEHPALPTASPGLQDSWEWGGCSPDMGFGERFSKDFLDSREPHRDIHA

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 belongs 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*).

RMRLHNN*