Supplemental Data

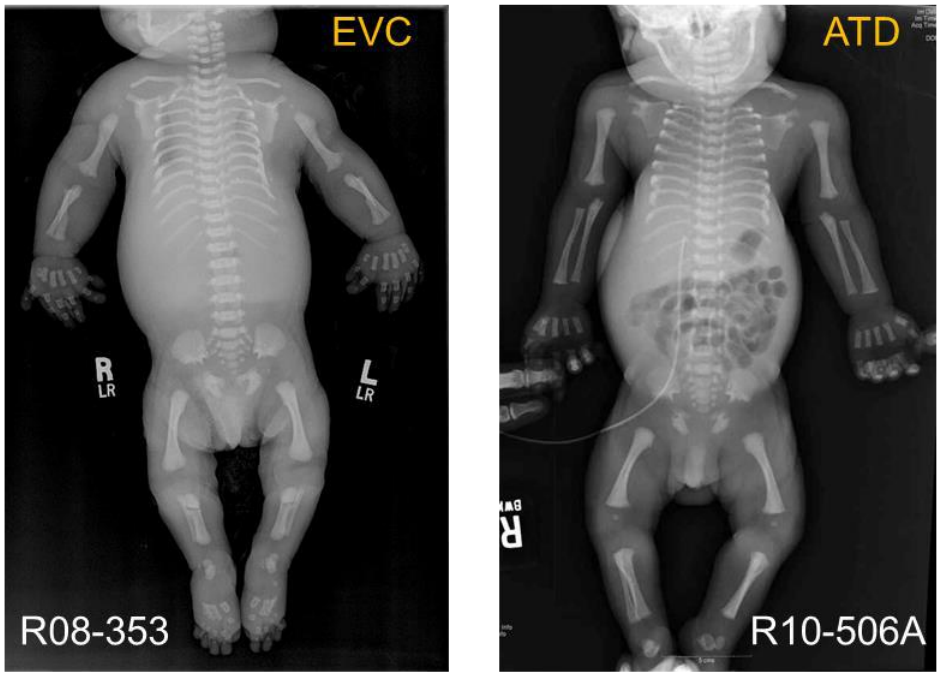

Figure S1. Representative radiographs for EVC and ATD. In the EVC case (R10-506A, neonatal, *EVC2* mutation), the chest is narrow with relatively well developed ribs, there is slight reverse campomelia of the humeri along with polydactyly and well-ossified digits. The lower extremities are well-ossified but the fibulae are thin. There is accelerated ossification of the knee epiphyses. In the ATD case (R08-353, neonatal, *DYNC2H1*) note the long, narrow chest, moderately short ribs and short extremities. The ends of the long bones have metaphyseal abnormalities and there is a trident appearance to the pelvis. The overall skeletal findings are similar to all types of SRPS but with less severe skeletal involvement.

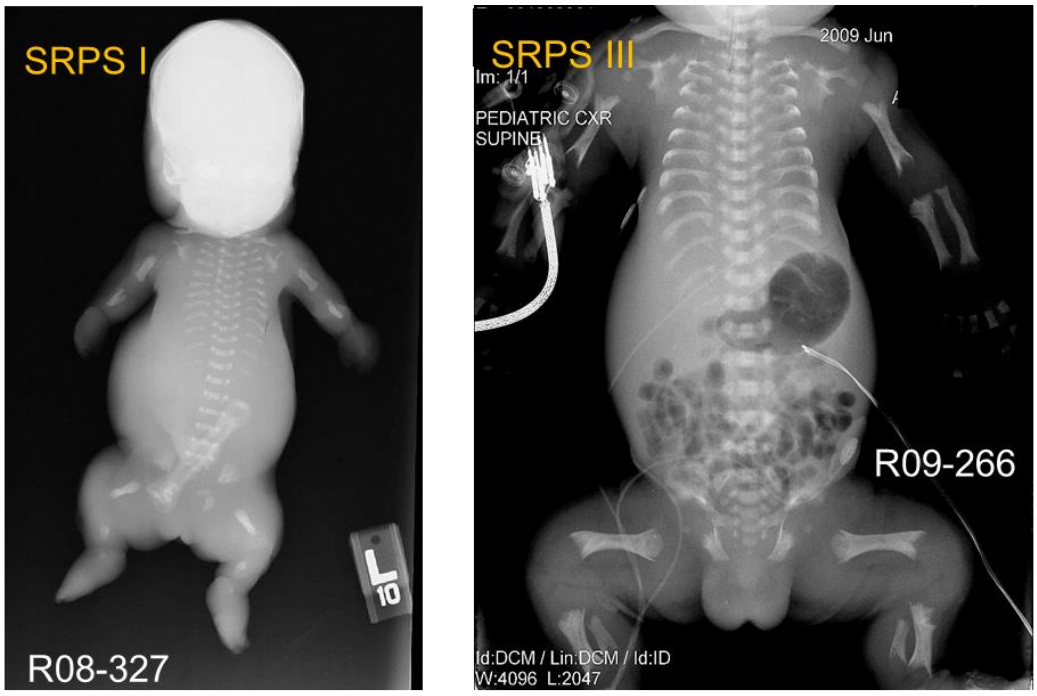

Figure S2. Representative radiographs for SRPS types I and III. For SRPS type I (R08-327, 28 weeks, *DYNC2H1*) there is extreme micromelia and very short, poorly mineralized long bones. The chest is very small, with very short ribs and hypoplastic scapulae. Polydactyly is typically present. For SRPS type III (R09-266, neonatal, *DYNC2H1*) the radiographic abnormalities are less severe but small chests and poorly formed scapulae are typical. The long bones are short and there are metaphyseal abnormalities with lateral spikes at the ends. Ilia are small and a trident pelvis is common. Polydactyly is a variable feature.

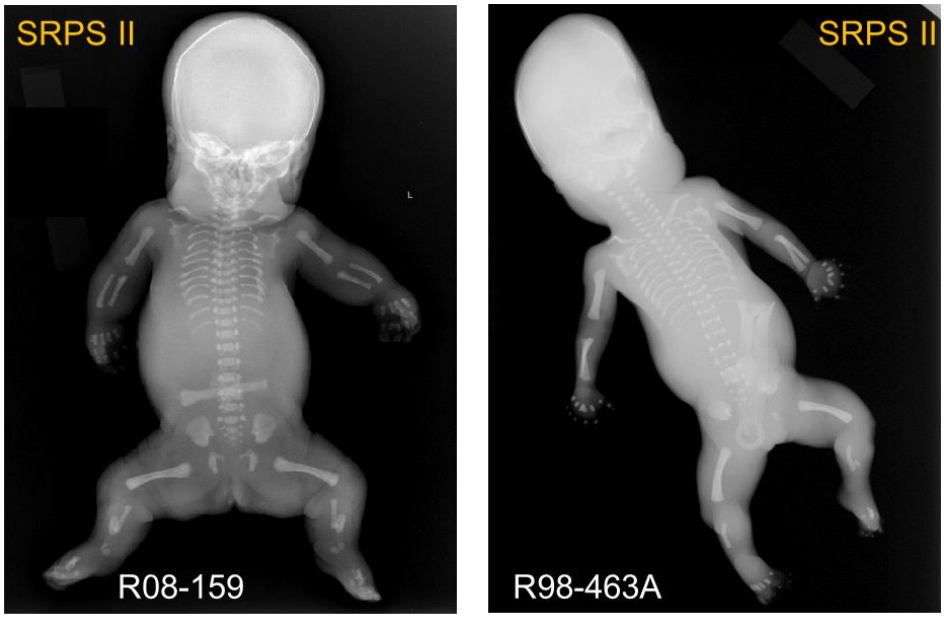

Figure S3. Representative radiographs for SRPS type II. In both individuals (R08-159, neonatal, *NEK1*; R98-463A, 16 weeks, *NEK1*) there are small chests with hypoplastic long bones that have smooth ends. Fibulae are small and rounded. Polydactyly was universally present.

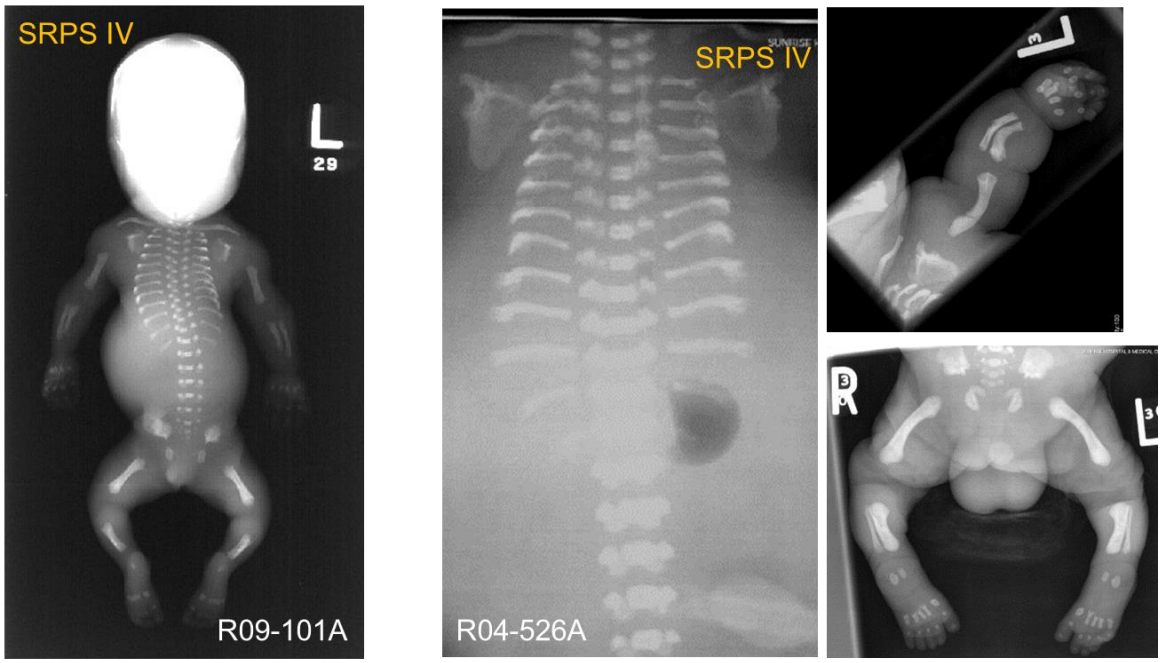

Figure S4. Representative radiographs for SRPS type IV. In both individuals (R09-101A, 21 weeks, *IFT80*; R04-526A, neonatal, *WDR19*) there are small chests with horizontal ribs and short, bent long bones with smooth ends. Fibulae are thin. Polydactyly was absent.

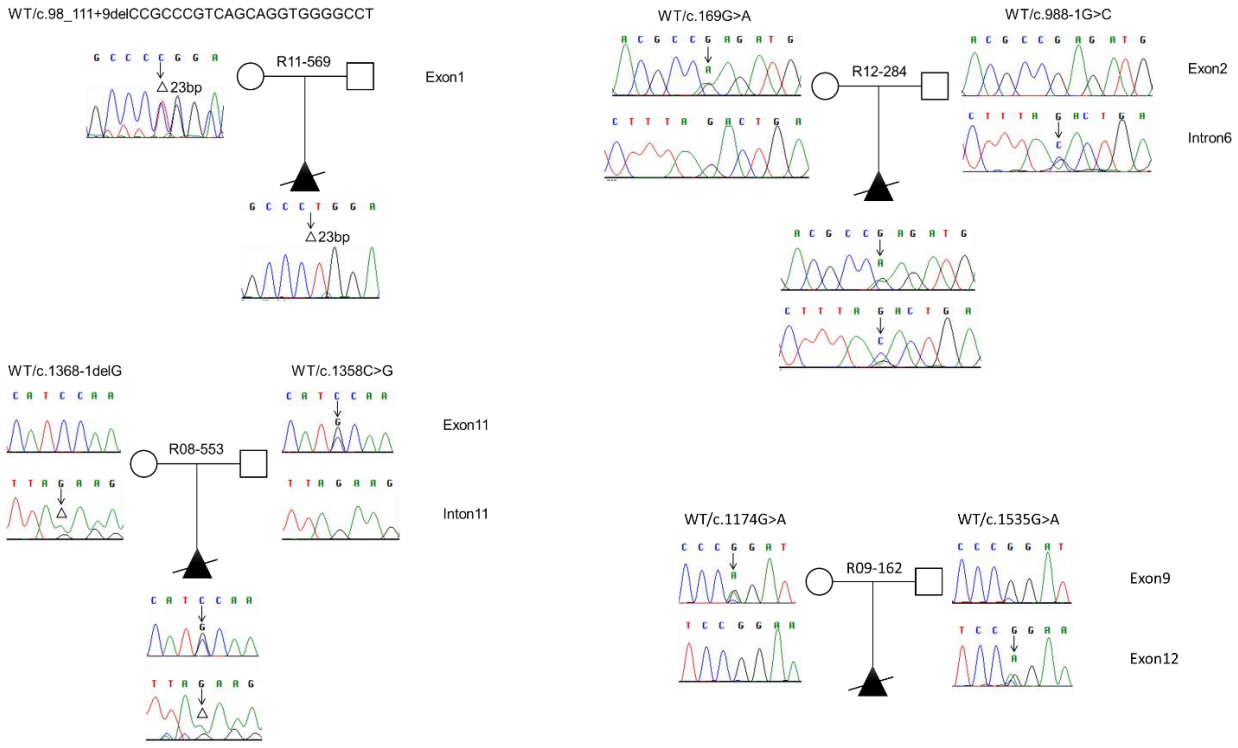

Figure S5. Families with *FUZ, TRAF3IP1* and *LBR* mutations. Sanger sequence confirmation of the mutations in each family is shown on the pedigree. The reference sequence is shown above and black arrows identify the locations of the mutations (R11-569, *FUZ*; R08-553, *TRAF3IP1*; R12-284, *TRAF3IP1*; R09-162, *LBR*).