Figure S2. D17S1174 analysis in 200 Japanese control subjects, showing discontinuous distribution of the CA repeat numbers, as observed in the Japanese families with limb malformations.

A. Distribution of CA repeat numbers. Although a slippage phenomenon would account for the CA repeat numbers of 19 and 18, it does not explain the discontinuous distribution.

B. Schematic representation of the mechanisms leading to the discontinuous distribution. Unequal interchromosomal/interchromatid exchanges affecting the CA repeat stretch can lead to non-continuous extension and shortening of the CA repeat number, and intrachromatid rearrangements affecting the CA repeat stretch can result in non-continuous shortening of the CA repeat number.