



**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.