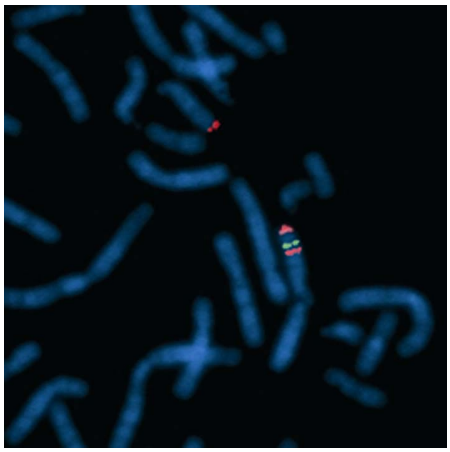
LEGENDS TO SUPPLEMENTARY FIGURES

Supplementary Figure 1. FISH confirmation of a hemizygous deletion of 15q24 in patient C45/06, using a 15q subtelomere probe (red), and BAC probes (green/red) specific to the deletion region. FISH using the same probes in the mother of patient C45/06 did not provide any evidence for paracentric inversion of the deletion region, as has been shown to occur in the transmitting parent for a number of other recurrent genomic disorders (2,10-16) (data not shown). FISH confirmation of the deletions in patients ID204, IMR349 and IMR371 has been published previously (2,3).

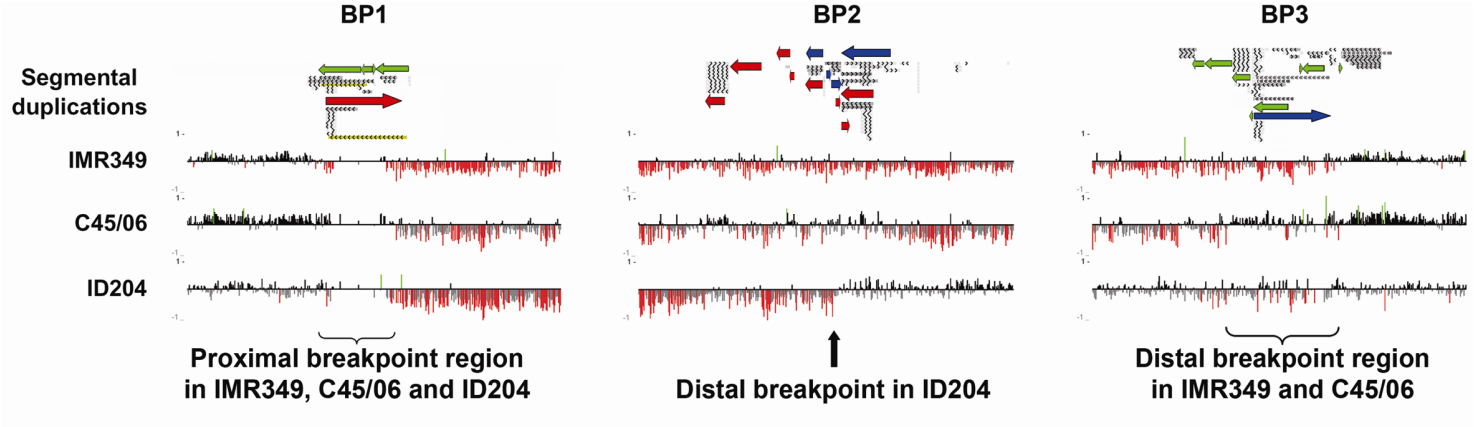
Supplementary Figure 2. Breakpoint mapping of three 15q24 deletions. Each panel shows oligonucleotide array data for a 250 kb region surrounding BP1, BP2 and BP3 (BP1, chr15:72,050,000-72,300,000; BP2, chr15:73,715,000-73,965,000; BP3, chr15:75,880,000-76,130,000). For each individual, deviations of probe $\log_2$ ratios from zero are depicted by grey/black bars, with those exceeding a threshold of 1.5 standard deviations from the mean probe ratio colored green and red to represent relative gains and losses, respectively. Tracks above each plot indicate segmental duplications (grey/yellow/orange bars representing duplications with 90-98%/98-99%/99-100% sequence identity, respectively). Pairwise segmental duplication relationships between each breakpoint region are represented by colored arrows (identity to BP1 in blue, identity to BP2 in green, identity to BP3 in red). Due to the presence of copy number polymorphism at BP1 (4,5) and cross-hybridization of probes within each segmental duplication cluster, the exact extent of the deletions are difficult to define, but in each case they localize to the segmental duplications shared between each breakpoint.

Supplementary Figure 3. Additional images showing facial and digital features of (**A**-**C**) patient ID204 as an adult, and (**D**, **E**) patient C45/06.

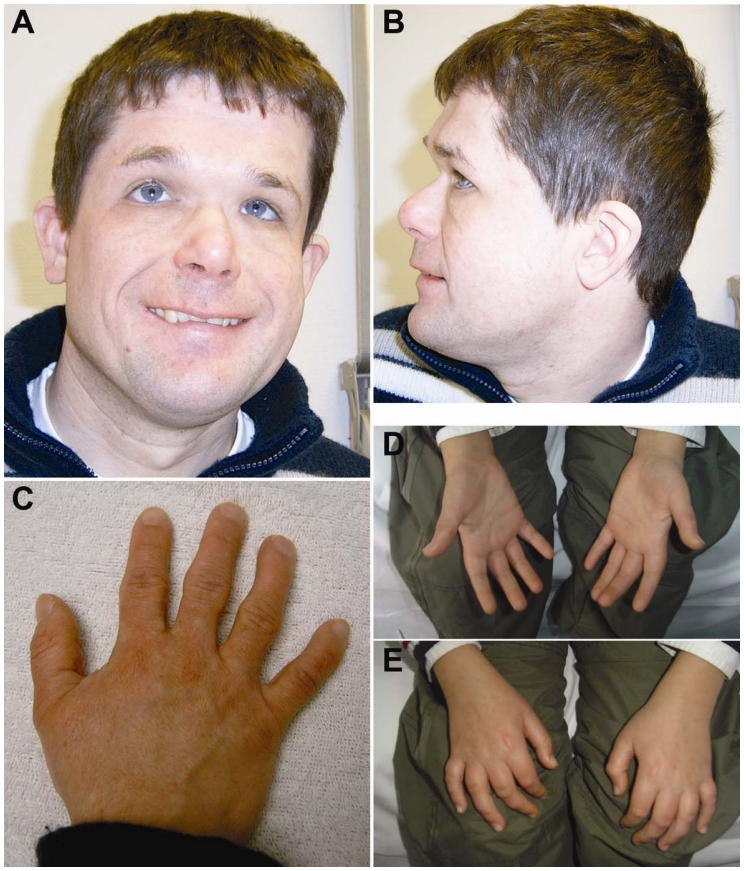
Supplementary Figure 4. Sagittal brain MRI scans of patient C45/06 showing (**A**) thin corpus callosum and hypoplasia of the anterior pituitary gland and (**B**) ectopic posterior pituitary enhanced after gadolinium administration.



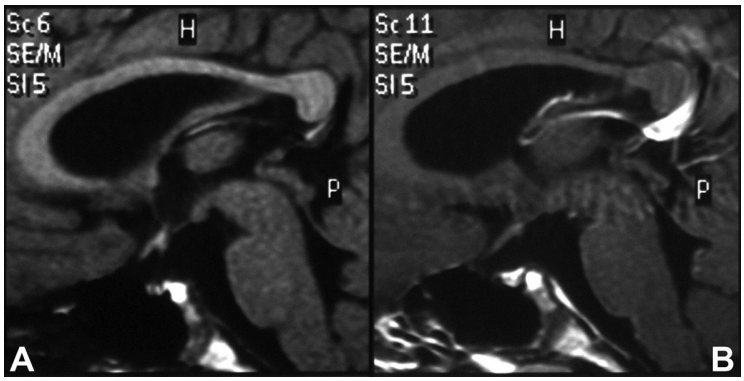
Supplementary Figure 1



Supplementary Figure 2



Supplementary Figure 3



Supplementary Figure 4