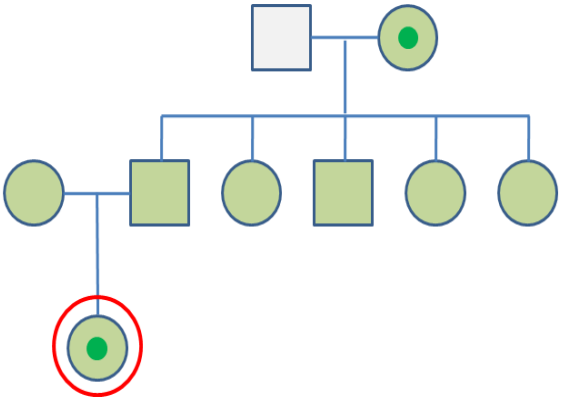
Supplementary Material 4: Microsatellite analysis

Genotypes from three microsatellite markers within the duplicated region were available from a previous study (unpublished study conducted at deCODE genetics) for one of the identified carriers (Ctrl 1) as well as for many of her close relatives. The markers are D15S128 (Chr15:22,681,798-22,682,092), D15S97 (Chr15:24,382,615-24,382,758), and D15S156 (Chr15:25,580,600-25,580,939) see also figure 1 in main article. Genotypes were available for Ctrl 1, both her parents, four of her father's siblings, and her paternal grandmother (supplementary figure 2). One of the three microsatellites, D15S156, is not very polymorphic within the family so it was relatively uninformative. We therefore present only the results for the other two microsatellites, D15S97 and D15S128, although it should be noted that the observed alleles of D15S156 are consistent with these results. The genotype trace-files are shown in supplementary figures 3a and 3c for D15S97 and D15S128, respectively; and the allele calls are laid on top of the trace-files in supplementary figures 3b and 3d for the respective markers (for genotyping methods see Kong *et al*. (1)).

Supplementary figure 2: Extended family with microsatellite genotypes at 15q11-q13

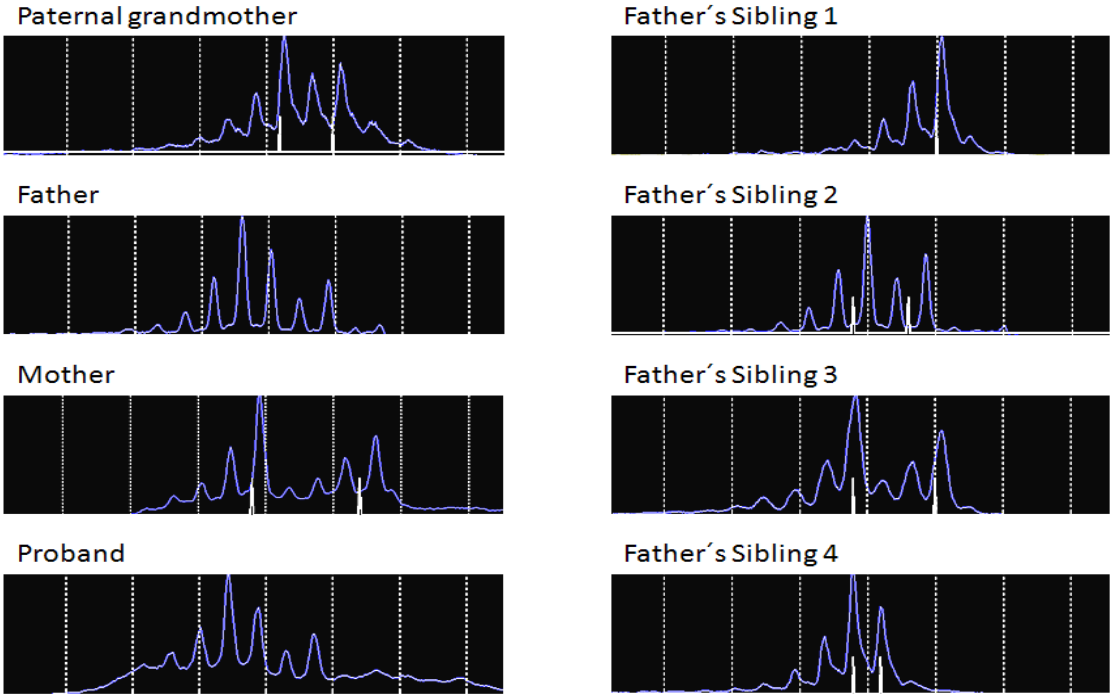


Red circle indicates Ctrl 1, who was identified in the SGENE+ dataset. Family members from which microsatellite genotypes are available are indicated with light green. In addition to Ctrl 1, her paternal grandmother is also genotyped on the HumanCNV370 microarray (dark green dot).

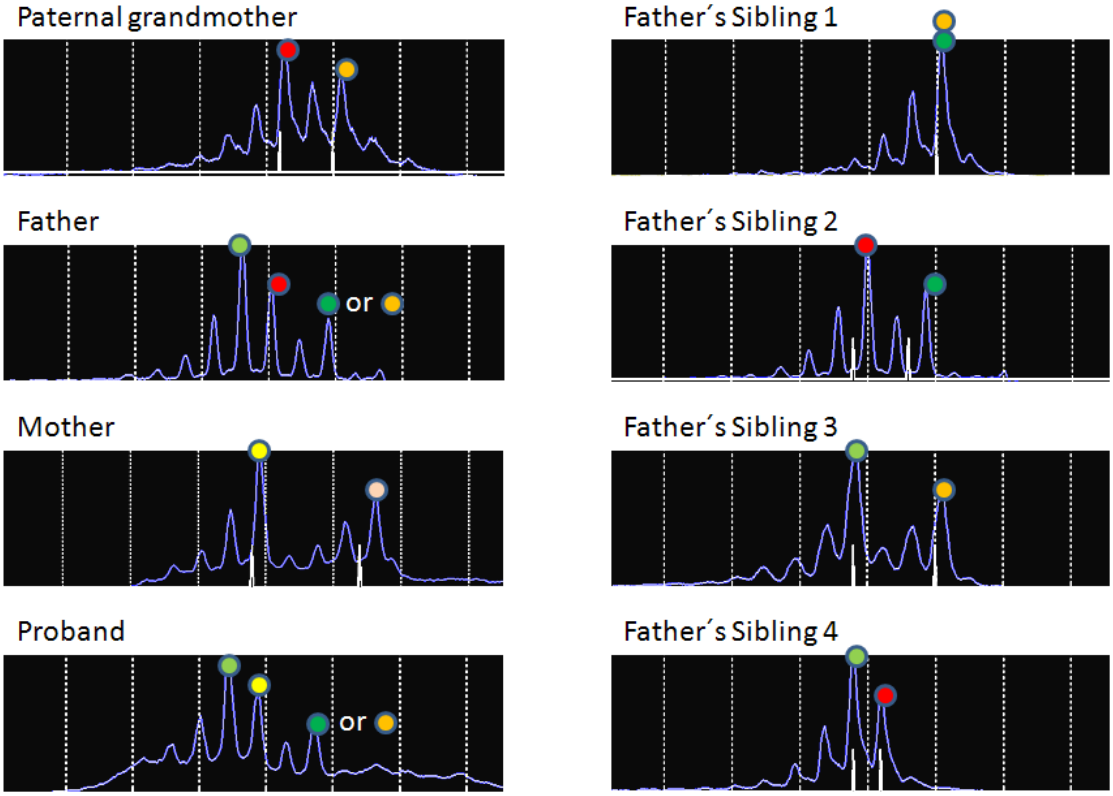
From the microarray data we already know that Ctrl 1 carries the duplication whereas the paternal grandmother does not. From the traces of D15S97 (supplementary figures 3a-b) we see that the father of Ctrl 1 carries the duplication and has passed it on to Ctrl 1. As the paternal grandparents have one allele in common (the grandfather's alleles can be inferred from his children's alleles), we cannot determine the duplication origin in the father. None of the father's four siblings carries the duplication, which must therefore be *de novo* in the father, since his four siblings carry single copies of both alleles from the grandfather. For D15S128 (supplementary figures 3c-d) the paternal grandparents are double heterozygotes, and we can determine that the *de novo* duplication in the father involves both chromosomes of his father, indicating inter-chromosomal non-allelic homologous recombination as the most likely mechanism.

1. Kong A, Gudbjartsson DF, Sainz J, Jonsdottir GM, Gudjonsson SA, Richardsson B, Sigurdardottir S, Barnard J, Hallbeck B, Masson G, Shlien A, Palsson ST, Frigge ML, Thorgeirsson TE, Gulcher JR, Stefansson K: A high-resolution recombination map of the human genome. Nat Genet 2002; 31:241-247

Supplementary figure 3a: View of genotype trace files for D15S97 in the extended family.

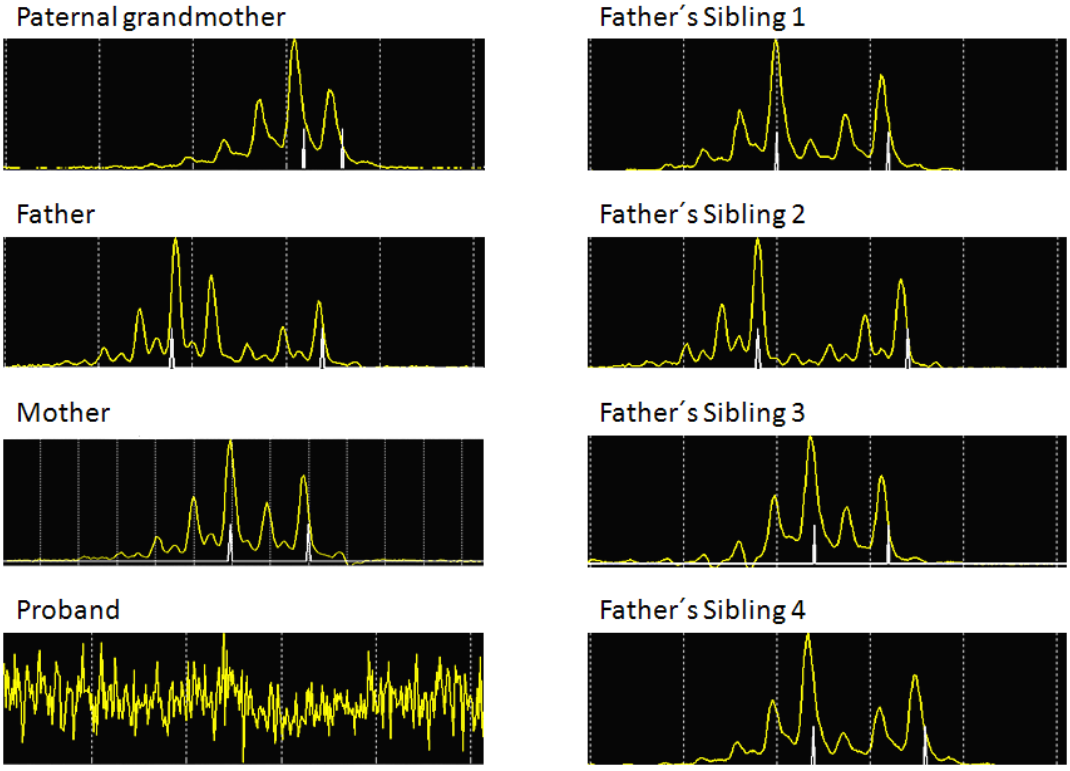


Supplementary figure 3b: View of genotype trace files for D15S97 with allele calls overlaid.

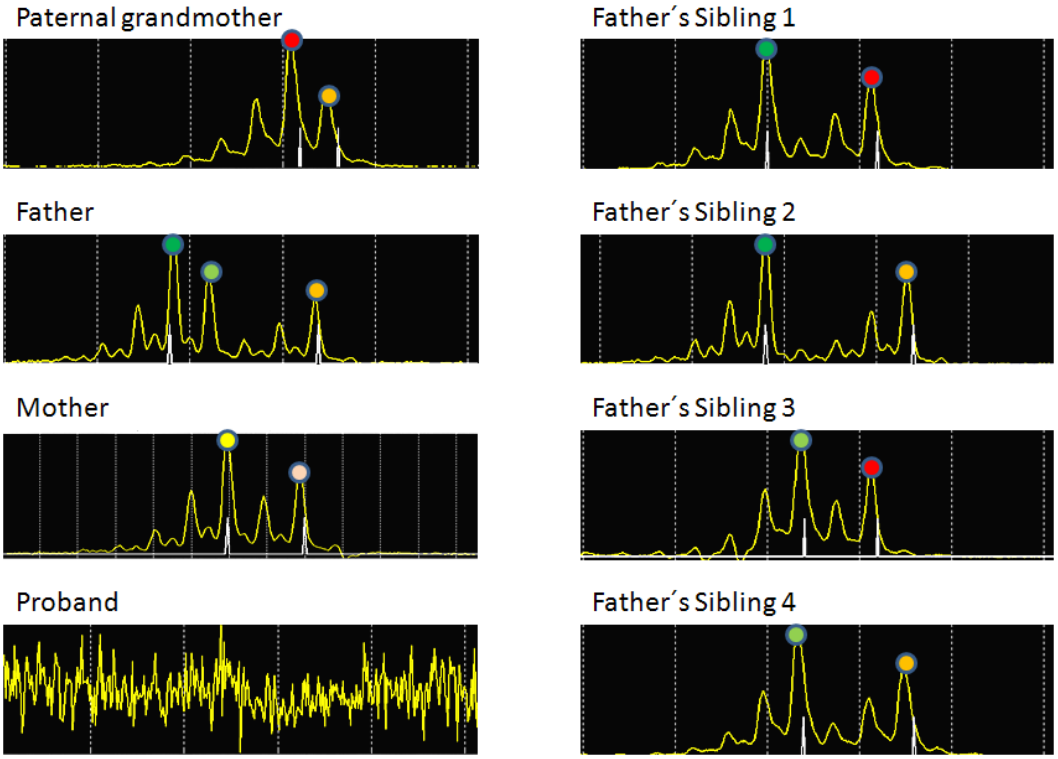


Red and orange dots indicate the alleles of the paternal grandmother, dark and light green the alleles of the paternal grandfather (not genotyped), and yellow and beige the alleles of Ctrl 1's mother.

Supplementary figure 3c: View of genotype trace files for D15S128 in the extended family.



Supplementary figure 3d: View of genotype trace files for D15S128 with allele calls overlaid.



Red and orange dots indicate the alleles of the paternal grandmother, dark and light green the alleles of the paternal grandfather (not genotyped), and yellow and beige the alleles of Ctrl 1´s mother. The assay failed for Ctrl 1 for this microsatellite.