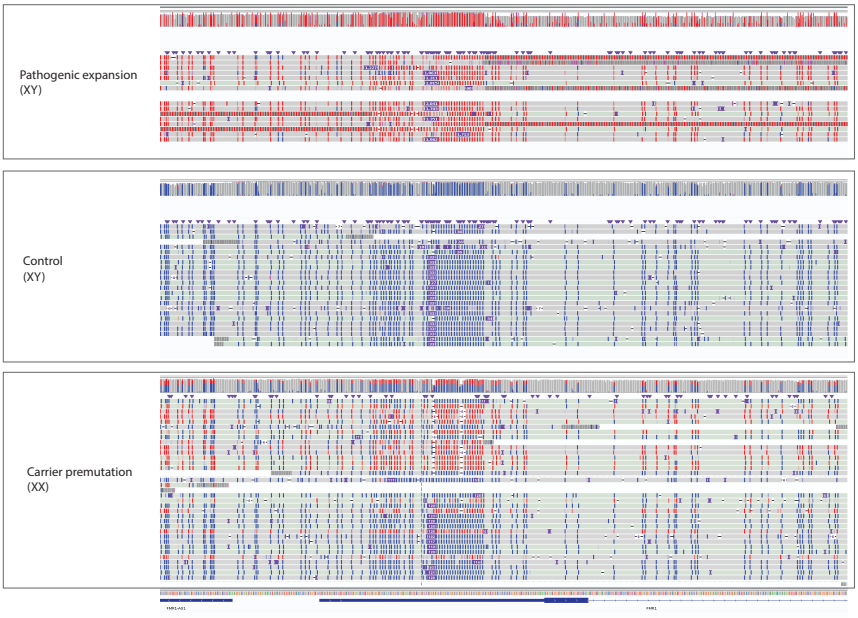

Fig. S1. Methylation pattern across *FMR1*. The integrative genomics viewer (IVG) screenshots showing the beginning and upstream region of *FMR1* with methylated CpGs in red and unmethylated CpGs in blue. Top: A male with a full expansion (highlighted by a purple box on the read) shows hypermethylation. Middle: A male control displays hypomethylation. Bottom: A female carrier of a premutation-range expansion, one allele is expanded (purple box) and the other is normal. The expanded allele is predominantly hypomethylated, while the normal allele is predominantly hypermethylated.

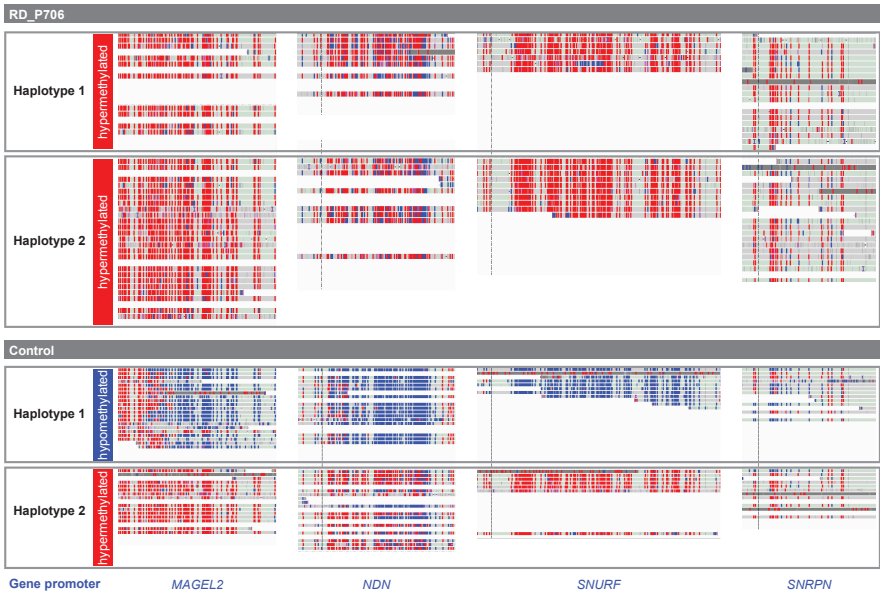

Fig. S2. Methylation pattern across promoters of genes in the Prader-Willi region. Integrative genomics viewer (IVG) screenshots with phased reads spanning the promoters of MAGEL2, NDN, SNURF and SNRPN, showing methylated CpGs in red and unmethylated CpGs in blue. The individual with maternal uniparental disomy of chromosome 15 (top) shows hypermethylation across all four gene promoters on both haplotypes. In a control individual (bottom), the methylation pattern shows haplotype 1 as hypomethylated (paternal) and haplotype 2 as hypermethylated (maternal).

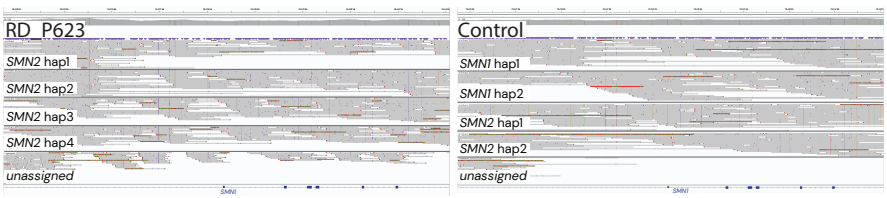

Fig. S3. Phasing of *SMN1* and *SMN2*. Integrative genomics viewer (IVG) screenshot across the *SMN1* region. In individual with homozygous deletion of *SMN1* (RD_P623), no haplotypes corresponding to *SMN1* are observed, while four haplotypes correspond to *SMN2*. In the control individual, there are two haplotypes identified as *SMN1* and two as *SMN2*.

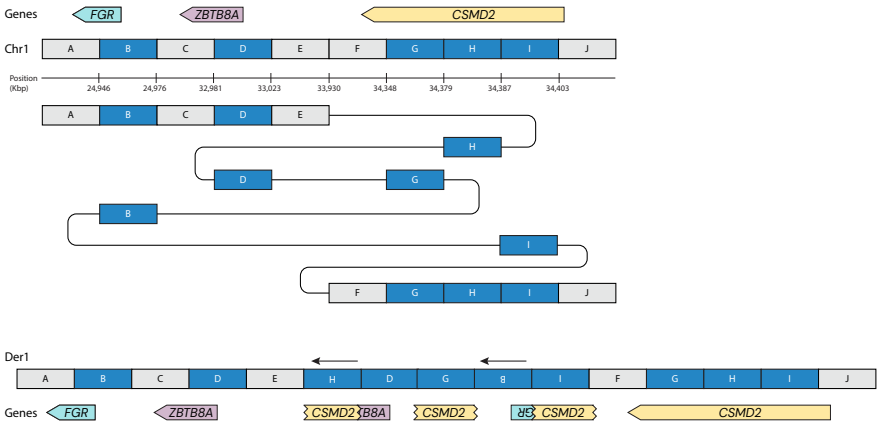

Fig. S4. Chromoanasynthesis event of chromosome 1p. Schematic of the reference chromosome 1 across the affected region, with duplicated segments shown in blue, and genes at breakpoints displayed above. Middle: Subway plot of the rearrangement, illustrating duplicated segments inserted between segments E and F in an disorganized manner. Bottom: Resolved derivative structure, with inverted segments indicated by arrows. Below, a depiction of the genes involved in the rearrangement.

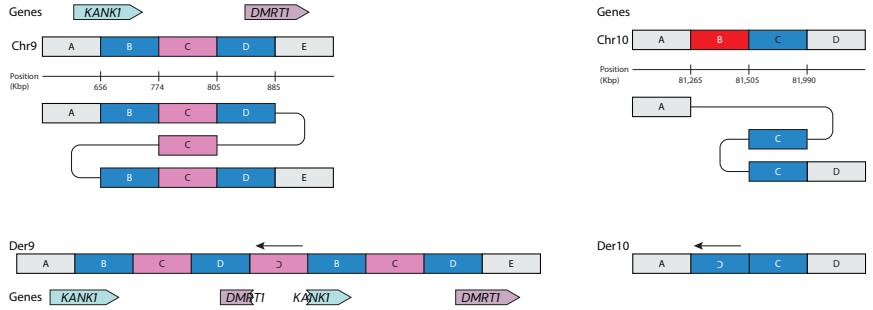

Fig. S5. Complex SVs on chromosomes 9p and 10p. Top: Schematic of the reference chromosome 9 and 10 across the affected region, with deleted segments (red), duplicated segments (blue) and triplicated segments (pink). Genes at breakpoints displayed above. Middle: Subway plot of the rearrangements, showing a DUP-INV/TRIP-DUP and DEL-INV-DUP structure, respectively. Bottom: Resolved derivative structures with inverted segments indicated by arrows. Below, a depiction of the genes involved in the rearrangement.

Commented [ME1]: I do not have the resolved structure yet.