**Short legend for figures:**

x axis - position on chromosome

y axis - zygosity (# of alternative variant reads / # of total reads in particular genomic position)

orange lines - position of centromere (UCSC hg19)

red horizontal line - theoretical line separating heterozygous and homozygous

red continuous line - moving average of zygosity, window 2000 variants; when it reaches 1, region could be a loss of heterozygosity (LOH), potential recessive disease hotspot

blue dots - SNVs in their positions

green dots - rare, nonsynonymous coding variants, according to FunSeq2 (prematureStop + nonsynonymous)

orange dots - rare, nonsynonymous coding variants present only in Zimmer sample (not in Synder/Giab)

**Reference :**  
Stokowy, T, et al. (2016) RareVariantVis: new tool for visualization of causative variants in rare monogenic disorders using whole genome sequencing data. Bioinformatics btw359