



**Supplementary Figure S1. Results showing the integrated approaches (array-CGH and targeted-WES) performed on the Italian POI cohort.** Among the 41 patients of the cohort, 17 were subjected to the combined method (Bestetti et al., 2019), and the results are shown in the enlarged Venn diagram. For each patient, the obtained data are shown, and the intersection shows the patients positive for both rare ovary-related CNVs and SNVs/Indels. CGH, comparative genomic hybridisation; CNV, copy number variant; FDe, full deletion; FDU, full duplication; fs, frameshift; NR, never reported variant; PDe, partial deletion; PDU, partial duplication; POI, primary ovarian insufficiency; PPE, potential position effect; SNVs, single-nucleotide variants; WES, whole exome sequencing.