**S3 Table: HSCR disease gene characteristics**



*Most known HSCR genes are intolerant to genetic variation and are rarely impacted by CNVs in unaffected individuals[1, 2]. These genes have been described to be impacted by CNVs in HSCR patients[3, 4], although this does not seem to be a frequent phenomenon, as in our cohort we did not detect any CNV impacting a known HSCR gene. Abbreviations: mis\_z: Missense variation Z-score, syn\_z:* *Synonymous variation z-sore****,*** *PLI****:*** *probability of being loss-of-function intolerant , PRec: probability of being tolerant to heterozygous loss of function variation but intolerant to homozygous loss of function variation, PNull: probability of being tolerant to loss of function variation (heterozygous or homozygous), del: deletion, dup: duplication, EW: embryonic week, DDD: deciphering developmental disorders project. Data derived from (https://gnomad.broadinstitute.org/) and the DDD control track at: https://genome-euro.ucsc.edu/*

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