Solve-RD: systematic pan-European data sharing and collaborative analysis to solve rare diseases

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Solve-RD analysis projects for existing exome and genome data

Title	DITF	WG
Solve-RD QC analysis: Perform quality control on all Solve-RD samples	All	Several WGs
Solve-RD ancestry and admixture analysis: For all Solve-RD samples, determine ancestry and admixture	All	Several WGs
Solve-RD variant annotation: Annotate all Solve-RD samples using multiple databases	All	WG1 SNV/indel
Automated SNV and Indel filtering and prioritisation	All	WG1 SNV/indel
Prevalence of pathogenic variant in genes associated in with known tumour risk syndromes	GENTURIS	WG1 SNV/indel & WG5 Meta-analysis
Detection of mitochondrial DNA variants from WES/WGS	All	WG1 SNV/indel
Landscape of rare genetic variants in titin gene	EURO NMD	WG1 SNV/indel
Clinvar class IV-V mutational burden analysis of exome negative (unsolved) patients with intellectual disability (ID)	ITHACA	WG1 SNV/indel & WG5 Meta-analysis
Detection of Copy Number Variants in WES and WGS data experiment in the Solve-RD re-analysis cohort	All	WG2 CNV
Solve-RD CNV analysis using Conifer: detect possible copy-number variations using multiple CNV tools	All	WG2 CNV
Identification and Interpretation of rare structural variants in WES-based rare-disease diagnostics	All	WG2 CNV
Solve-RD STR analysis: detect aberrant short tandem repeats	All	WG2 CNV
Solve-RD UPD analysis: detect possible uniparental disomies	All	WG2 CNV
Run of homozygosity, consanguinity, relatedness and ancestry analysis	All	WG3 RoH / relatedness
Solve-RD de novo variant calling in patient-parent trios	All	WG4 *De novo* mutations
Solve-RD meta-analysis: Compare case and control cohorts to find novel disease genes based on statistical enrichment and overlap analysis	All	WG5 Meta-analysis