Materials List for:

Targeted Next-generation Sequencing and Bioinformatics Pipeline to Evaluate Genetic Determinants of Constitutional Disease

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Materials

Name	Company	Catalog Number	Comments
4 mL EDTA K2 tubes	Fisher Scientific	02-689-4	
1 M Tris Buffer	Bio Basic Canada Inc.	SD8141	
Gentra Puregene Blood Kit	Qiagen	158389	1,000 mL Kit. This is the blood extraction kit, referred to in step 1.3.
NanoDrop-1000 Spectrophotometer	Thermo Fisher Scientific	ND-2000	Replaced by the NanoDrop-2000 Spectrophotometer. This is the full-spectrum spectrophotometer, referred to in steps 1.4 and 2.1.2.
Qubit 2.0 fluorometer	Invitrogen	Q32866	This is a fluorometer appropriate for the quantification of DNA, referred to in steps 2.1.4, 2.1.6, 2.2.3, and 3.1.3.
Nextera Rapid Custom Capture Enrichment Kit	Illumina, Inc.	FC-140-1009	Specifically designed for the ONDRISeq panel, sequencing the exons of 80 genes, resulting in 971,388 base pairs of sequence in paired-end reads of 150 bases in length; 288 samples per kit. This is the target enrichment kit, referred to in steps 2.2, 2.2.2, 2.2.3, 3.1.5, 3.1.6, 3.4.1, and the Discussion.
2100 BioAnalyzer	Agilent Technologies	G2939BA	This is a automated electrophoresis system, referred to in step 3.1.4.

High Sensitivity DNA Reagent Kit	Agilent Technologies	5067-4626	110 Samples per kit; This is a DNA quality analysis kit, referred to in step 3.1.4.
MiSeq Reagent Kit v3	Illumina, Inc.	MS-102-3003	600 Cycle Kit; This is the NGS desktop instrument reagent kit, referred to in step 3.1.
MiSeq Personal Genome Sequencer	Illumina, Inc.	SY-410-1003	This is a NGS desktop instrument, referred to in steps 2.2.1, 3.1, 3.1.1, 3.1.2, 3.1.8, 3.2, 4.2.6, the Representative Results, and the Discussion.
Experiment Manager	Illumina, Inc.		This is NGS technology software, referred to in step 3.1.1 and **Figure 1**. https://support.illumina.com/sequencing/sequencing_software/experiment_manager/downloads.html
BaseSpace	Illumina, Inc.	SW-410-1000	This is a cloud-based computing environment, referred to in steps 3.1.2, 3.2, 3.3, 3.3.1, 3.3.2, 3.4, 3.4.1, 3.4.2 and 3.4.3. https://basespace.illumina.com/
CLC Genomics Workbench 10.1.1	Qiagen	832000	Open source options for data pre-processing are also available that can model the workflow used in this protocol. This is the software used for data pre-processing, referred to throughout step 4 and in **Figure 2**.
Annotate Variation			http://annovar.openbioinformatics.org/en/latest/user-guide/download/
RefSeq	National Center for Biotechnology Information		https://www.ncbi.nlm.nih.gov/refseq/
dbSNP138	National Center for Biotechnology Information		https://www.ncbi.nlm.nih.gov/projects/SNP/snp_summary.cgi?view+summary=view+summary&build_id=138
Exome Aggregation Consortium	Broad Institute		http://exac.broadinstitute.org/
National Heart, Lung, and Blood Institute Exome Sequencing Project European Cohort	University of Washington and the Broad Institute		http://evs.gs.washington.edu/EVS/
ClinVar	National Center for Biotechnology Information		https://www.ncbi.nlm.nih.gov/clinvar/
Combined Annotation Dependent Depletion	University of Washington and Hudson-Alpha Institute for Biotechnology		http://cadd.gs.washington.edu/
Sorting Intolerant from Tolerant	J. Craig Venter Instutite		http://sift.jcvi.org/
PolyPhen-2	Brigham and Women's Hospital, Harvard Medical School		http://genetics.bwh.harvard.edu/pph2/
Human Gene Mutation Database	Qiagen	834050	This is a disease mutation database, referred to in step 5.2 and the Representative Results. https://portal.biobase-international.com/cgi-bin/portal/login.cgi?redirect_url=/hgmd/pro/start.php
Splicing-based Analysis of Variants	Frey lab, University of Toronto		http://tools.genes.toronto.edu/
Human Splicing Finder	Aix Marseille Université		http://www.umd.be/HSF3/HSF.shtml

Other materials			
Centrifuge			
Disposable transfer pipets			