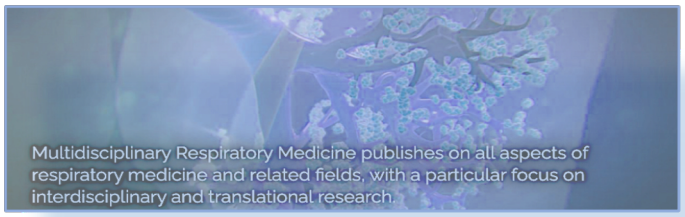

SUPPLEMENTARY MATERIAL

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Germline variant of *CTC1* gene in a patient with pulmonary fibrosis and myelodysplastic syndrome

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METHODS

Whole exome sequencing (WES)

Blood samples from the patient were collected and processed for genomic DNA isolation using MagCore® Genomic DNA Whole Blood Kit (RBC Bioscience, USA). Whole exome library was processed using the KAPA Hyper Prep Kit, HyperExome probes, and HyperCap Enrichment Kit and Bead Kit (Roche, USA) according to the SeqCap EZ HyperCap Workflow v3.2 following the recommended protocols. Paired-end 2x75 bp sequencing was performed on the Illumina NextSeq 500 Sequencer (Illumina Inc., USA). The raw sequencing reads were aligned to the GRCh38 (hg18) human reference genome using the BWA-mem algorithm, version 0.7.15, PCR duplicates were identified with the MarkDuplicates tool from Picard. Germline single nucleotide variants (SNV) and indels were detected by the GATK HaplotypeCaller. Annotation of obtained SNV/indels was performed with Annovar. To identify clinically relevant findings, only WES variants with total coverage of at least 10x, minor allele frequency (MAF) values in the non-Finnish European population ≤0.01, and predicted possible-probable deleteriousness were included. To predict the pathogenic significance of the emerging variants, mutation-prediction tools (SIFT, REVEL, PolyPhen-2, MutationTaster, and Align GVGD) were used. The segregation of *CTC1* variant c.1360delG was determined by Sanger sequencing using a BigDye Terminator v3.1 Cycle Sequencing Kit according to the manufacturer´s protocol. The primers were designed for the detection of exon 8 in the *CTC1* gene (F-primer: CGGTTAACTCTGCCTGGGTT, R-primer: AGATCCAGGGTAGGAGCCAG). Capillary sequencing was performed using BigDye-terminator chemistry on a 3500 Genetic Analyzer (Applied Biosystems, USA).

Supplementary Table 1. Virtual genes panel associated with myeloid malignancies and predispositions to myeloid and pulmonary disorders.

ACD	*DBA6*	*HAX1*	*RECQL3*	*SCKL1*
BGS	*DBA7*	*HK1*	*RECQL4*	*SF3B1*
ALDH2	*DBA8*	*CHEK2*	*RFWD3*	*SH2B3*
ANKRD26	*DDX41*	*IKZF1*	*RPL11*	*SIDBA1*
ASXL1	*DKC1*	*LIG4*	*RPL15*	*SIDBA2*
ATG2B	*DKCX*	*MAD2L2*	*RPL23*	*SIDBA3*
ATM	*ELANE*	*MLASA1*	*RPL26*	*SIDBA4*
ATP11C	*EPCAM*	*MLH1*	*RPL36*	*SIFD*
BLM	*ERCC4*	*MPL*	*RPL5*	*SLC25A38*
BRCA1	*ETV6*	*MSH2*	*RPS10*	*SLX4*
BRCA2	*FAAP100*	*MSH6*	*RPS15*	*SRP72*
BRIP1	*FAN1*	*MYH9*	*RPS17*	*STN1*
BTK	*FANCA*	*NAF1*	*RPS19*	*STRA13*
C19ORF40	*FANCB*	*NBS1*	*RPS24*	*TERC*
C1ORF86	*FANCC*	*NF1*	*RPS26*	*TERT*
CBL	*FANCD2*	*NHP2*	*RPS27A*	*TET2*
CDAN1	*FANCE*	*NOP10*	*RPS28*	*TINF2*
CDAN2	*FANCF*	*PALB2*	*RPS29*	*TMPRSS6*
CDAN3	*FANCG*	*PARN*	*RPS7*	*TP53*
CDAN4	*FANCI*	*PAX5*	*RPL27*	*TRMA*
CEBPA	*FANCL*	*PMS2*	*RPL31*	*TSR2*
CTC1	*FANCM*	*PTPN11*	*RPL35A*	*TTP*
DBA1	*G6PC3*	*RAD51*	*RTEL1*	*UBE2T*
DBA2	*GATA1*	*RAD51C*	*RUNX1*	*WAS*
DBA3	*GATA2*	*RBBP6*	*SAMD9*	*WRAP53*
DBA4	*GP*	*RBM8A*	*SAMD9L*	*XRCC2*
DBA5	*GSKIP*	*RECQL2*	*SBDS*	*XRCC6*
				ZBTB32