Table 3 Variants identified in the whole exome sequencing of the IPF cohort

Case no	Unique features	Age at onset	Gene (Transcript No)	Location	Nucleotide and protein change	Type	Zygosity	Classification	Inheritance	Disease	In silico prediction				Frequency in database			
											SIFT	LRT	Polyphen	Mutation taster 2	1000 genome	GnomAD (V2.1)	GnomAD (V3.1)	Med Var
Case1	Current smoker, third degree consanguinity, CPFE	68	***TERT*** (ENST00000310581.10)	Exon 15	c.3208G>A (p.Val1070Met)	Missense	Heterozygous	Uncertain significance (PM2, PP3)	Autosomal dominant	Pulmonary fibrosis and/or bone marrow failure syndrome, telomere related, 1 (OMIM#614742)	Damaging	-	Possibly Damaging	-	NA	NA	0.0006%	0.001%
Case 2	Never smoker, post COVID pulmonary sequelae, IHD with LV systolic dysfunction	70	***TTR*** (ENST00000237014.8)	Exon 1	c.34G>C (p.Ala12Pro)	Missense	Heterozygous	Uncertain significance	Autosomal dominant	Hereditary transthyretin related Amyloidosis (OMIM#105210) Familial carpal tunnel syndrome (OMIM#115430)	Damaging	Damaging	Probably Damaging	Damaging	NA	NA	NA	0.000964%
Case 3	No cough, Reformed smoker, CPFE, post-tb pulmonary sequelae	*58*	***MUC5B*** (ENST00000529681.5)	Exon 22	c.3004G>A (p.Val1002Met)	Missense	Heterozygous	Uncertain significance	Autosomal dominant	Susceptibility to idiopathic pulmonary fibrosis (OMIM#178500)	Damaging	NA	Probably Damaging	Benign	NA	0.003%	0.006%	0.04%
Case 4	No cough, Stable disease, not on antifibrotics, Psoriasis, Pulmonary hypertension, post COVID pulmonary sequelae	*67*	***MUC5B*** *(ENST00000529681.5)*	Exon 31	c.11224T>G (p.Ser3742Ala)	Missense	Heterozygous	Uncertain significance	Autosomal dominant	Interstitial lung disease (OMIM#178500)	Tolerated	NA	Benign	Benign	0.019%	0.006%	0.001%	0.055%
Case 5	Never smoker Post COVID pulmonary sequelae	*57*	***CFAP74*** ***(***ENST00000682832.2***)***	Exon 24 Novel	c.2810G>C (p.Arg937Thr)	Missense	Heterozygous	Uncertain significance	Autosomal recessive	Primary ciliary dyskinesia-49 without situs inversus (OMIM#620197)	Damaging	NA	Benign	NA	NA	NA	NA	0.009%

				Exon 18	c.2020A>G (p.Ser674Gly)	Missense	Heterozygous										0.01%	NA
Case 6	IHD, slow progression of IPF over 7 years, coexisting COPD	*63*	***DSP*** (ENST00000379802.8)	Exon 23	c.4687_4688del (p.Leu1563GlufsTer63)	Frameshift	Heterozygous	Likely pathogenic	Autosomal dominant	Arrhythmogenic right ventricular dysplasia - 8 (OMIM#607450)	NA	NA	NA	Damaging	NA	NA	NA	0.001%
Case 7	Early onset, reformed smoker	43	***ECT2*** (ENST00000392692.8)	Exon 16 Novel	c.1616A>G (p.Tyr539Cys)	Missense	Heterozygous	Uncertain significance	-	-	Damaging	Damaging	Probably Damaging	Damaging	NA	NA	NA	NA