Supplemental Table S1: Characteristics of the Genetic Mutations and Number of ILD Individuals Included from Each Kindred

Gene	DNA Change*	Impact on Protein*	Frequency in ExAC database†	Number of ILD patients with Blood Telomere Length data	Telomere length, ln Rel(T/S) (± S.D.)	Age-adjusted Telomere Length (± S.D.)	Telomere length percentile (mean)	References (other studies)	Number of Individuals Included in this study
TERT	c.97C>T	p.Pro33Ser	Absent	1	0.855	-0.69	<1	[1, 2]	1
TERT	c.293C>A	p.Ala98Asp	Absent	1	0.450	-1.06	<1		1
TERT	c.377C>A	p.Thr126Lys	Absent	1	1.167	-0.42	2-3	[3]	1
TERT	c.416T>G	p.Leu139Arg	Absent	1	0.843	-0.81	<1		1
TERT	c.430G>A	p.Val144Met	Absent	3	0.95±0.03	-0.61±0.02	<1	[1, 2]	5
TERT	c.569C>T	p.Ala190Val	0.00001765	1	1.235	-0.47	1-2		1
TERT	c.1002_1004delCTC	p.Ser335del	Absent	1	0.909	-0.55	<1	[3]	1
TERT	c.1397G>C	p.Arg466Pro	Absent	1	0.817	-0.70	<1		1
TERT	c.1417G>C	p.Val473Leu	Absent	1	1.137	-0.39	3-4		1
TERT	c.1456C>T	p.Arg486Cys	Absent	2	1.03±0.01	-0.46±0.11	1-2	[1-3]	2
TERT	c.1603C>T	p.Arg535Cys	Absent	1	0.739	-0.68	<1		1
TERT	c.1710G>T	p.Lys570Asn	Absent	1	0.735	-0.78	<1	[3, 4]	1
TERT	c.1892G>A	p.Arg631Gln	Absent	4	0.75±0.23	-0.75±0.25	<1	[2]	4
TERT	c.1895C>T	p.Pro632Leu	Absent	1	0.231	-1.37	<1		1
TERT	c.2006C>T	p.Arg669Gln	Absent	1	1.197	-0.31	9-10		1
TERT	c.2011C>T	p.Arg671Trp	Absent	1	0.813	-0.60	<1	[2]	1
TERT	c.2033C>A	p.Ala678Asp	Absent	1	0.659	-0.96	<1	[5]	1
TERT	c.2080G>A	p.Val694Met	Absent	1	1.113	-0.41	2-3	[2, 6, 7]	1
TERT	c.2105C>T	p.Pro702Leu	Absent	3	1.03±0.36	-0.51±0.35	<1	[2, 3, 6]	4
TERT	c.2110C>T	p.Pro704Ser	Absent	3	1.12±0.16	-0.36±0.15	4-5	[2, 6, 8, 9]	3
TERT	c.2240delT	p.Val747fsX19	Absent	2	1.04±0.25	-0.46	1-2	[1, 2]	4
TERT	c.2266C>T	p.Arg756Cys	0.000008238	1	0.832	-0.79	<1		1
TERT	c.[2431C>T;2433C>T]‡	p.Arg811Cys	Absent	2	0.57±0.40	-0.89±0.38	<1		2
TERT	c.2469-2A>T	Splicing	Absent	2	0.72±0.22	-0.87±0.23	<1		2
TERT	c.2521C>T	p.Leu841Phe	Absent	1	1.090	-0.42	2-3	[10]	1
TERT	c.2539G>A	p.Gly847Ser	Absent	1	1.247	-0.35	6-7	[3]	1

TERT	c.2572C>T	p.Arg858Trp	Absent	1	0.829	-0.66	<1		1
TERT	c.2593C>T	p.Arg865Cys	Absent	1	1.031	-0.47	1-2	[1-3]	1
TERT	c.2594G>A	p.Arg865His	0.00001278	2	1.01±0.17	-0.54±0.18	<1	[1-3]	5
TERT	c.2599G>A	p.Val867Met	Absent	1	1.148	-0.36	5-6	[2]	1
TERT	c.2621C>G	p.Thr874Arg	Absent	2	0.83±0.35	-0.66±0.37	<1		2
TERT	c.2647T>A	p.Phe883Ile	Absent	1	1.083	-0.50	<1		1
TERT	c.2775C>A	p.His925Gln	0.00001674	1	1.615	0.06	50-51§	[2]	2
TERT	c.2851C>T	p.Arg951Trp	0.000008290	2	1.14±0.22	-0.36±0.15	5-6	[2]	4
TERT	c.2869A>C	p.Ser957Arg	Absent	1	0.580	-0.95	<1	[2, 6]	1
TERT	c.2912G>A	p.Arg971His	Absent	1	0.925	-0.59	<1		1
TERT	c.2947C>T	p.His983Tyr	Absent	1	1.077	-0.50	<1		1
TERT	c.3055C>T	p.Leu1019Phe	Absent	1	0.926	-0.57	<1	[2, 6]	2
TERT	c.3148A>G	p.Lys1050Glu	Absent	1	1.057	-0.47	1-2	[2, 6]	1
TERT	c.3150G>C	p.Lys1050Asn	0.00008867	1	1.064	-0.42	2-3		1
TERT	c.3187G>A	p.Gly1063Ser	Absent	2	1.21±0.69	-0.33±0.67	6-7	[2]	3
TERT	c.3202G>A	p.Glu1068Lys	Absent	2	0.83±0.21	-0.66±0.21	<1	[3]	2
TERT	c.3346_3522del177	p.Glu1116fsX11	Absent	1	1.073	-0.50	<1	[1, 2]	2
TERC	r.11g>t		Absent	1	1.236	-0.41	3-4		1
TERC	r.37a>g		0.00001827	1	1.047	-0.45	1-2	[1, 11]	1
TERC	r.66c>a		Absent	1	0.615	-1.05	<1		1
TERC	r.116c>t		Absent	1	0.862	-0.67	<1	[12, 13]	1
TERC	r.182g>c		Absent	2	0.61±0.06	-0.89±0.03	<1	[3]	2
TERC	r.234c>g		Absent	1	1.058	-0.56	<1		1
RTEL1	c.602delG	p.Gly201GlufsX15	Absent	1	1.205	-0.27	15-16	[14]	2
RTEL1	c.1451C>T	p.Pro484Leu	Absent	1	1.008	-0.55	<1	[14]	5
RTEL1	c.1940C>T	p.Pro647Leu	Absent	1	1.118	-0.43	2-3	[14]	1
RTEL1	c.2005C>T	p.Gln669X	Absent	1	0.871	-0.64	<1	[14]	2
RTEL1	c.2063C>G	p.Ser688Cys	0.000008572	1	1.028	-0.48	1-2		1
RTEL1	c.2205_2207delGAC	p.Asp736del	Absent	1	0.814	-0.66	<1		2
RTEL1	c.3371A>C	p.His1124Pro	Absent	1	1.062	-0.53	<1	[14]	1
PARN	c.246-2A>G	Splicing	0.00001394	2	0.99±0.04	-0.47±0.06	1-2	[14]	5
PARN	c.529C>T	p.Gln177X	Absent	1	0.957	-0.67	<1	[14]	4

PARN	c.563_564insT	p.Ile188IlefsX7	Absent	1	1.297	-0.22	24-25	[14]	2
PARN	c.751delA	p.Arg251GlufsX14	Absent	1	1.196	-0.29	12-13	[14]	1
PARN	c.874delG	p.Asp292ThrfsX16	Absent	2	1.16±0.14	-0.30±0.05	10-11		3
PARN	c.1081+1G>A	Splicing	Absent	1	1.245	-0.19	30-31	[14]	2
PARN	c.1262A>G	p.Lys421Arg	0.000008294	1	1.083	-0.37	4-5	[14]	1
PARN	c.1749_1750delAG	p.Glu585AspfsX4	Absent	1	1.129	-0.35	6-7		1

*The positions of the DNA, RNA and protein variants are described using TERT NM_198253.2 (isoform 1), TERC NR_001566.1, PARN NM_002582.3 (isoform 1) and RTEL1 NM_001283009.1 (isoform 3).

†The frequency (if present) of the variants found in the Exome Aggregation Consortium (ExAC) database version 0.3 (www.exac.broadinstitute.org) are listed.

‡Both variants are found in two siblings, suggesting that they in *cis* and in the same allele.

§This same rare variant was found by directly sequencing lung tissue from an affected sister with IPF, liver dysfunction, leukopenia, and anemia. Telomerase activity as measured by the *in vitro* telomere repeat amplification protocol (TRAP) was 40.4%[2].

Supplemental Table S2: Selected Comorbid Conditions of ILD Subjects with Heterozygous *TERT, TERC, RTEL1* or *PARN* Mutations

	Total (N=115)	***TERT* (N=75)**	***TERC* (N=7)**	***RTEL1* (N=14)**	***PARN* (N=19)**	**P-value**
Musculoskeletal, N (%)						
Osteopenia	8 (7.0)	6 (8.0)	1 (14.3)	1 (7.1)	0	0.45
Osteoporosis	12 (10.4)	6 (8.0)	1 (14.3)	0	5 (26.3)	0.054
Scoliosis	2 (1.7)	1 (1.3)	0	1 (7.1)	0	0.36
Psychiatry, N (%)						
Depression	15 (13)	11 (14.7)	0	2 (14.3)	2 (10.5)	0.86
Anxiety	7 (6.0)	4 (4.0)	2 (28.6)	1 (7.1)	0	0.080
Endocrine, N (%)						
Hypothyroidism	17 (14.8)	8 (10.7)	1 (14.3)	3 (21.4)	5 (26.3)	0.26
Diabetes Mellitus	8 (7.0)	3 (4.0)	0	1 (7.1)	4 (21.1)	0.070
Cardiovascular, N (%)						
Atherosclerosis	11 (9.6)	5 (6.7)	0	4 (28.6)	2 (28.6)	0.082
CHF	3 (2.6)	2 (2.7)	0	0	1 (5.3)	0.73

The prevalence of comorbid conditions is compared using Fisher's exact test.

Abbreviations used: CHF, congestive heart disease

Supplemental Table S3: Radiographic Characteristics of ILD Subjects with Heterozygous *TERT, TERC, RTEL1* or *PARN* Mutations

	Total	*TERT*	*TERC*	*RTEL1*	*PARN*	P-value
Number Subjects, N	73	52	4	10	7	
Overall Diagnosis						
Definite UIP	34 (46.6)	25 (48.1)	1 (25.0)	6 (60.0)	2 (28.6)	0.54
Possible UIP	24 (32.9)	17 (32.7)	2 (50.0)	1 (10.0)	4 (57.1)	0.16
Inconsistent with UIP	15 (20.5)	10 (19.2)	1 (25.0)	3 (30.0)	1 (14.3)	0.78
Radiographic Feature, N	73	52	4	10	7	
Reticulation	72 (98.6)	52 (100)	4 (100)	9 (90.0)	7 (100)	0.29
Intralobular Septal Thickening	71 (97.3)	51 (98.1)	4 (100)	9 (90.0)	7 (100)	0.50
Macrocystic Honeycombing	37 (50.7)	26 (50)	2 (50.0)	5 (50.0)	4 (57.1)	1
Microcystic Honeycombing	44 (60.3)	33 (63.5)	3 (75.0)	3 (30.0)	5 (71.4)	0.20
Traction Bronchiectasis	71 (97.3)	52 (100)	4 (100)	8 (80.0)	7 (100)	0.053
Nodules	15 (20.5)	11 (21.2)	1 (25.0)	2 (20.0)	1 (14.3)	1
Parenchymal Bands	22 (30.1)	17 (32.7)	2 (50.0)	3 (30.0)	0	0.23
Predominant Ground Glass Opacities*	7 (9.6)	6 (11.5)	1 (25.0)	0	0	0.41
Emphysema	10 (13.7)	4 (7.7)	1 (25.0)	4 (40.0)	1 (14.3)	**0.032**
Apical-Basal Distribution, N	73	52	4	10	7	
Upper Lobe Predominant	6 (8.2)	4 (7.7)	0	2 (20.0)	0	0.51
Lower Lobe Predominant	46 (63.0)	33 (63.5)	2 (50.0)	6 (60.0)	5 (71.4)	0.91
Diffuse	21 (28.8)	15 (28.8)	2 (50.0)	2 (20.0)	2 (28.6)	0.71
Axial Distribution, N	73	52	4	10	7	
Subpleural	57 (78.1)	41 (78.8)	2 (50.0)	8 (80.0)	6 (85.7)	0.56
Peribronchovascular	0	0	0	0	0	-
Diffuse	16 (21.9)	11 (21.2)	2 (50.0)	2 (20.0)	1 (14.3)	0.56
Symmetry, N	73	52	4	10	7	
Right > Left	14 (19.2)	10 (19.2)	0	2 (20.0)	2 (28.6)	0.85
Left > Right	9 (12.3)	8 (15.4)	0	0	1 (14.3)	0.58
Symmetric	50 (68.5)	34 (64.5)	4 (100)	8 (80.0)	4 (57.1)	0.47
Air-trapping, N	60	40	4	10	6	
None	27 (45.0)	19 (47.5)	3 (75.0)	5 (50.0)	0	0.075
Mild	20 (33.3)	13 (32.5)	0	4 (40.0)	3 (50.0)	0.45
Moderate	10 (16.7)	7 (17.5)	0	1 (10.0)	2 (33.3)	0.63
Severe	3 (5.0)	1 (2.5)	1 (25.0)	0	1 (16.7)	0.086
Pulmonary Artery Size, N	70	49	4	10	7	
< 3 cm	39 (55.7)	28 (57.1)	3 (75.0)	4 (40.0)	4 (57.1)	0.69
3-3.5 cm	25 (35.7)	17 (34.7)	1 (25.0)	5 (50.0)	2 (28.6)	0.78
> 3.5 cm	6 (8.6)	4 (8.2)	0	1 (10.0)	1 (14.3)	0.85
Mediastinal and Hilar Lymph Node Size, N	70	49	4	10	7	
< 1 cm	35 (50)	27 (55.1)	1 (25.0)	3 (30.0)	4 (57.1)	0.36

1-1.5 cm	23 (32.9)	16 (32.7)	3 (75.0)	2 (20.0)	2 (28.6)	0.30
> 1.5 cm	12 (17.1)	6 (12.2)	0	5 (50.0)	1 (14.3)	**0.045**
Pleural Fat, N	63	42	4	10	7	
None	4 (6.3)	1 (2.4)	1 (25.0)	2 (20.0)	0	0.056
Mild	36 (57.1)	23 (54.8)	2 (50.0)	6 (60.0)	5 (71.4)	0.89
Marked	23 (36.5)	18 (42.9)	1 (25.0)	2 (20.0)	2 (28.6)	0.59

*Predominant Ground Glass Opacities refers to ground glass that is out of proportion of degree of fibrosis, or separated from areas of fibrosis.

The data shown are N (%). The prevalence of radiographic characteristics is compared using Fisher's exact test. P-values < 0.05 are shown in bold.

Supplemental Table S4: Factors Associated with the Presence of Emphysema in ILD Subjects with Heterozygous *TERT, TERC, RTEL1* or *PARN* Mutations

	Emphysema Present (n=10)	Emphysema Absent (n=63)	P-value
Male, N (%)	8 (80%)	34 (54%)	0.17
Age, mean (SD)	59.9 (5.8)	55.0 (13.2)	0.14
Smoking, N (%)	8 (80%)	23 (36.5%)	0.015
Gene group, N (%)			
TERT	4 (40.0%)	48 (76.2%)	0.028
TERC	1 (10.0%)	3 (4.8%)	0.45
RTEL1	4 (40.0%)	6 (9.5%)	0.026
PARN	1 (10.0%)	6 (9.5%)	1

Unadjusted P-values were calculated using Fisher's exact test (categorical variables) or Wilcoxon rank-sum test (age at diagnosis).

Supplemental Table S5: Histopathologic Characteristics of ILD Subjects with Heterozygous *TERT, TERC, RTEL1* or *PARN* Mutations

	All (N=42)	***TERT* (N=32)**	***TERC* (N=2)**	***RTEL1* (N=6)**	***PARN* (N=2)**	**P-value**
Histopathologic Diagnosis, N (%)						
UIP	21 (50.0)	16 (50.0)	1 (50.0)	2 (33.3)	2 (100)	0.62
NSIP	4 (9.5)	3 (9.4)	0	1 (16.7)	0	0.68
DIP	1 (2.4)	1 (3.1)	0	0	0	1
PPFE	5 (11.9)	3 (9.4)	1 (50.0)	1 (16.7)	0	0.25
Unclassifiable	3 (7.1)	2 (6.2)	0	1 (16.7)	0	0.57
Organizing DAD	1 (2.4)	1 (3.1)	0	0	0	1
SRIF	4 (9.5)	3 (9.4)	0	1 (16.7)	0	0.68
Chronic HP	2 (4.7)	2 (6.2)	0	0	0	1
> 1 Pattern*	1 (2.4)	1 (3.1)	0	0	0	1
Honeycombing, N (%)						
Mild	11 (26.2)	10 (31.3)	0	1 (16.7)	0	0.90
Moderate	10 (23.8)	7 (21.9)	2 (100)	1 (16.7)	0	0.13
Severe	12 (28.6)	9 (28.1)	0	1 (16.7)	2 (100)	0.19
Fibroblastic Foci, N (%)						
Mild	12 (28.6)	10 (31.3)	0	2 (33.3)	0	1
Moderate	7 (16.7)	6 (18.8)	0	1 (16.7)	0	1
Severe	15 (35.7)	12 (37.5)	1 (50.0)	0	2 (100)	0.060
Interstitial Fibrosis, N (%)						
Mild	1 (2.4)	0	0	1 (16.7)	0	0.24
Moderate	5 (11.9)	4 (12.5)	0	1 (16.7)	0	1
Severe	36 (85.7)	28 (87.5)	2 (100)	4 (66.7)	2 (100)	0.60
Peribronchovascular Fibrosis, N (%)						
Mild	14 (33.3)	11 (34.4)	0	3 (50.0)	0	0.56
Moderate	13 (31.0)	9 (28.1)	2 (100)	1 (16.7)	1 (50.0)	0.15
Severe	10 (23.8)	9 (28.1)	0	1 (16.7)	0	1
Hyalinized or Ropey Collagen, N (%)						
Mild	10 (23.8)	8 (25.0)	0	1 (16.7)	1 (50.0)	0.78
Moderate	5 (11.9)	5 (15.6)	0	0	0	0.75
Severe	5 (11.9)	3 (9.4)	0	2 (33.3)	0	0.51
Fibroelastosis, N (%)						
Mild	0	0	0	0	0	-
Moderate	2 (4.8)	2 (6.2)	0	0	0	1
Severe	5 (11.9)	3 (9.4)	1 (50.0)	1 (16.7)	0	0.25
Acute DAD, N (%)						
Mild	3 (7.1)	1 (3.1)	1 (50.0)	0	1 (50.0)	**0.027**
Moderate	1 (2.4)	1 (3.1)	0	0	0	1

Severe	1 (2.4)	1 (3.1)	0	0	0	1
Organizing DAD, N (%)						
Mild	2 (4.8)	2 (6.2)	0	0	0	1
Moderate	4 (9.5)	3 (9.4)	1 (50.0)	0	0	0.41
Severe	3 (7.1)	2 (6.2)	0	0	1 (50.0)	0.31
Acute Fibrinous Organizing Pneumonia, N (%)						
Mild	7 (16.7)	5 (15.6)	0	1 (16.7)	1 (50.0)	0.56
Moderate	0	0	0	0	0	-
Severe	0	0	0	0	0	-
Interstitial Inflammation, N (%)						
Mild	21 (50.0)	17 (53.1)	0	4 (66.7)	0	0.26
Moderate	14 (33.3)	11 (34.4)	2 (100)	0	1 (50.0)	**0.048**
Severe	4 (9.5)	2 (6.2)	0	1 (16.7)	1 (50.0)	0.17
Peribronchovascular Inflammation, N (%)						
Mild	14 (33.3)	10 (31.2)	1 (50.0)	3 (50.0)	0	0.56
Moderate	2 (4.8)	1 (3.1)	1 (50.0)	0	0	0.20
Severe	0	0	0	0	0	-
Granulomatous Inflammation, N (%)						
Loose Granulomas	1 (2.4)	1 (3.1)	0	0	0	1
Compact Granulomas	2 (4.8)	1 (3.1)	0	1 (16.7)	0	0.42
Lymphoid Aggregates, N (%)						
Mild	12 (28.6)	8 (25.0)	0	3 (50.0)	1 (50.0)	0.38
Moderate	6 (14.3)	5 (15.6)	0	0	1 (50.0)	0.33
Severe	0	0	0	0	0	-
Organizing Pneumonia, N (%)						
Mild	1 (2.4)	1 (3.1)	0	0	0	1
Moderate	4 (9.5)	3 (9.4)	0	1 (16.7)	0	0.68
Severe	1 (2.4)	1 (3.1)	0	0	0	1
Pulmonary Arterial Medial Thickening, N (%)						
Mild	15 (35.7)	13 (40.6)	0	2 (33.3)	0	0.73
Moderate	16 (38.1)	10 (31.3)	2 (100)	2 (33.3)	2 (100)	0.08
Severe	3 (7.1)	3 (9.4)	0	0	0	1

*Histopathologic specimen with evidence of PPFE and NSIP

The prevalence of histopathologic characteristics is compared using Fisher's exact test. P-values < 0.05 are shown in bold.

Abbreviations used: UIP, usual interstitial pneumonia; NSIP, nonspecific interstitial pneumonia; DIP, desquamative interstitial pneumonia; PPFE, pleuroparenchymal fibroelastosis; DAD, diffuse alveolar damage; SRIF, smoking related interstitial fibrosis; HP, hypersensitivity pneumonitis.

Supplemental Table S6. Correlation between Radiographic and Histopathologic Diagnoses for ILD Subjects with Heterozygous *TERT, TERC, RTEL1* or *PARN* Mutations (n=34)

Radiographic Diagnosis	All	Definite UIP (N=18)	Possible UIP (N=8)	Inconsistent with UIP (N=8)
Histopathologic Diagnosis, N (%)				
UIP	18 (52.9)	11 (61.1)	3 (37.5)	4 (50.0)
NSIP	3 (8.8)	1 (5.6)	1 (12.5)	1 (12.5)
PPFE	3 (8.8)	1 (5.6)	1 (12.5)	1 (12.5)
Unclassifiable	3 (8.8)	0	2 (25.0)	1 (12.5)
Organizing DAD	1 (2.9)	0	0	1 (12.5)
SRIF	3 (8.8)	3 (16.7)	0	0
Chronic HP	2 (5.8)	1 (5.6)	1 (12.5)	0
> 1 Pattern*	1 (2.9)	1 (5.6)	0	0

*More than one histopathologic diagnosis included evidence of PPFE and NSIP

Abbreviations used: UIP, usual interstitial pneumonia; NSIP, nonspecific interstitial pneumonia; PPFE, pleuroparenchymal fibroelastosis; DAD, diffuse alveolar damage; SRIF, smoking related interstitial fibrosis; HP, hypersensitivity pneumonitis.

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