Respiratory Disorders

Gene panel

Gene panel information

Gene panel	**Respiratory Disorders**
Version	4
Total genes	179
Activation date	Friday 26 july 2024
Publisher	Center for Medical Genetics, Ghent

Genes

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
ABCA3	99.96 %	601615	Surfactant metabolism dysfunction, pulmonary, 3, 610921 (3), Autosomal recessive
ABCC8	99.98 %	600509	Diabetes mellitus, permanent neonatal 3, with or without neurologic features, 618857 (3), Autosomal recessive, Autosomal dominant; Diabetes mellitus, transient neonatal 2, 610374 (3); Diabetes mellitus, noninsulin-dependent, 125853 (3), Autosomal dominant; Hypoglycemia of infancy, leucine-sensitive, 240800 (3), Autosomal dominant; Hyperinsulinemic hypoglycemia, familial, 1, 256450 (3), Autosomal recessive, Autosomal dominant
ACD	100 %	609377	?Dyskeratosis congenita, autosomal recessive 7, 616553 (3), Autosomal recessive, Autosomal dominant; ?Dyskeratosis congenita, autosomal dominant 6, 616553 (3), Autosomal recessive, Autosomal dominant
ACVRL1	99.88 %	601284	Telangiectasia, hereditary hemorrhagic, type 2, 600376 (3), Autosomal dominant
AGR2	99.98 %	606358	Respiratory infections, recurrent, and failure to thrive with or without diarrhea, 620233 (3), Autosomal recessive
AP3B1	99.89 %	603401	Hermansky-Pudlak syndrome 2, 608233 (3), Autosomal recessive
AQP1	99.99 %	107776	[Aquaporin-1 deficiency], 110450 (3); [Blood group, Colton], 110450 (3)
ARHGEF1	99.97 %	601855	?Immunodeficiency 62, 618459 (3), Autosomal recessive
ASAH1	99.9 %	613468	Spinal muscular atrophy with progressive myoclonic epilepsy, 159950 (3), Autosomal recessive; Farber lipogranulomatosis, 228000 (3), Autosomal recessive
ASCL1	99.82 %	100790	*No OMIM phenotypes*
ATP13A3	99.84 %	610232	Pulmonary hypertension, primary, 5, 265400 (3), Autosomal recessive
ATP6AP1	100 %	300197	Immunodeficiency 47, 300972 (3), X-linked recessive
BDNF	100 %	113505	*No OMIM phenotypes*
BLOC1S3	100 %	609762	Hermansky-Pudlak syndrome 8, 614077 (3), Autosomal recessive
BLOC1S6	99.98 %	604310	?Hermansky-Pudlak syndrome 9, 614171 (3), Autosomal recessive
BMP10	99.96 %	608748	*No OMIM phenotypes*
BMPR1B	99.61 %	603248	Acromesomelic dysplasia 3, 609441 (3), Autosomal recessive; Brachydactyly, type A2, 112600 (3), Autosomal dominant; Brachydactyly, type A1, D, 616849 (3), Autosomal dominant
BMPR2	99.95 %	600799	Pulmonary hypertension, familial primary, 1, with or without HHT, 178600 (3), Autosomal dominant; Pulmonary hypertension, primary, fenfluramine or dexfenfluramine-associated, 178600 (3), Autosomal dominant; Pulmonary venoocclusive disease 1, 265450 (3), Autosomal dominant
CARD11	99.97 %	607210	B-cell expansion with NFKB and T-cell anergy, 616452 (3), Autosomal dominant; Immunodeficiency 11B with atopic dermatitis, 617638 (3), Autosomal dominant; Immunodeficiency 11A, 615206 (3), Autosomal recessive

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CAV1	99.97 %	601047	Lipodystrophy, congenital generalized, type 3, 612526 (3), Autosomal recessive; Pulmonary hypertension, primary, 3, 615343 (3), Autosomal dominant; Lipodystrophy, familial partial, type 7, 606721 (3), Autosomal dominant
CCDC39	99.74 %	613798	Ciliary dyskinesia, primary, 14, 613807 (3), Autosomal recessive
CCDC40	100 %	613799	Ciliary dyskinesia, primary, 15, 613808 (3), Autosomal recessive
CD19	99.98 %	107265	Immunodeficiency, common variable, 3, 613493 (3), Autosomal recessive
CD81	99.97 %	186845	Immunodeficiency, common variable, 6, 613496 (3), Autosomal recessive
CFAP300	99.47 %	618058	Ciliary dyskinesia, primary, 38, 618063 (3), Autosomal recessive
CFTR	99.45 %	602421	Cystic fibrosis, 219700 (3), Autosomal recessive; Sweat chloride elevation without CF (3); Congenital bilateral absence of vas deferens, 277180 (3), Autosomal recessive; {Pancreatitis, hereditary}, 167800 (3), Autosomal dominant; {Bronchiectasis with or without elevated sweat chloride 1, modifier of}, 211400 (3), Autosomal dominant; {Hypertrypsinemia, neonatal} (3)
CIITA	99.99 %	600005	{Rheumatoid arthritis, susceptibility to}, 180300 (3); Bare lymphocyte syndrome, type II, complementation group A, 209920 (3), Autosomal recessive
CLEC1A	99.97 %	606782	{Aspergillosis, susceptibility to}, 614079 (3)
CLEC7A	99.98 %	606264	Candidiasis, familial, 4, autosomal recessive, 613108 (3), Autosomal recessive; {Aspergillosis, susceptibility to}, 614079 (3)
COPA	99.61 %	601924	{Autoimmune interstitial lung, joint, and kidney disease}, 616414 (3), Autosomal dominant
CR2	99.97 %	120650	{Systemic lupus erythematosus, susceptibility to, 9}, 610927 (3); ?Immunodeficiency, common variable, 7, 614699 (3), Autosomal recessive
CSF2RA	93.86 %	306250	Surfactant metabolism dysfunction, pulmonary, 4, 300770 (3)
CSF2RB	100 %	138981	Surfactant metabolism dysfunction, pulmonary, 5, 614370 (3), Autosomal recessive
CTC1	100 %	613129	Cerebroretinal microangiopathy with calcifications and cysts, 612199 (3), Autosomal recessive
CTLA4	99.99 %	123890	Immune dysregulation with autoimmunity, immunodeficiency, and lymphoproliferation, 616100 (3), Autosomal dominant; {Diabetes mellitus, insulin-dependent, 12}, 601388 (3); {Celiac disease, susceptibility to, 3}, 609755 (3); {Hashimoto thyroiditis}, 140300 (3), Autosomal dominant; {Systemic lupus erythematosus, susceptibility to}, 152700 (3), Autosomal dominant
CTNNBL1	100 %	611537	?Immunodeficiency 99 with hypogammaglobulinemia and autoimmune cytopenias, 619846 (3), Autosomal recessive
DAW1	99.9 %	620279	*No OMIM phenotypes*
DKC1	99.59 %	300126	?Cataracts, hearing impairment, nephrotic syndrome, and enterocolitis 1, 301108 (3), X-linked dominant; Dyskeratosis congenita, X-linked, 305000 (3), X-linked recessive
DNAAF1	99.99 %	613190	Ciliary dyskinesia, primary, 13, 613193 (3), Autosomal recessive
DNAAF2	99.91 %	612517	Ciliary dyskinesia, primary, 10, 612518 (3), Autosomal recessive
DNAH1	99.98 %	603332	Spermatogenic failure 18, 617576 (3), Autosomal recessive; Ciliary dyskinesia, primary, 37, 617577 (3), Autosomal recessive
DNAH11	99.93 %	603339	Ciliary dyskinesia, primary, 7, with or without situs inversus, 611884 (3), Autosomal recessive
DNAH5	99.98 %	603335	Ciliary dyskinesia, primary, 3, with or without situs inversus, 608644 (3), Autosomal recessive
DNAH9	99.99 %	603330	Ciliary dyskinesia, primary, 40, 618300 (3), Autosomal recessive
DNAI1	99.92 %	604366	Ciliary dyskinesia, primary, 1, with or without situs inversus, 244400 (3), Autosomal recessive

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DNAI2	99.86 %	605483	Ciliary dyskinesia, primary, 9, with or without situs inversus, 612444 (3), Autosomal recessive
DNAL1	99.79 %	610062	Ciliary dyskinesia, primary, 16, 614017 (3), Autosomal recessive
DOCK8	99.86 %	611432	Hyper-IgE recurrent infection syndrome, autosomal recessive, 243700 (3), Autosomal recessive
DTNBP1	99.89 %	607145	Hermansky-Pudlak syndrome 7, 614076 (3), Autosomal recessive
EFEMP2	99.94 %	604633	Cutis laxa, autosomal recessive, type IB, 614437 (3), Autosomal recessive
EIF2AK4	99.97 %	609280	Pulmonary venoocclusive disease 2, 234810 (3), Autosomal recessive
ELMOD2	99.8 %	610196	*No OMIM phenotypes*
ELN	99.86 %	130160	Cutis laxa, autosomal dominant, 123700 (3), Autosomal dominant; Supravalvar aortic stenosis, 185500 (3), Autosomal dominant
ENG	100 %	131195	Telangiectasia, hereditary hemorrhagic, type 1, 187300 (3), Autosomal dominant
EPHB4	99.82 %	600011	Capillary malformation-arteriovenous malformation 2, 618196 (3), Autosomal dominant; Lymphatic malformation 7, 617300 (3), Autosomal dominant
ERBIN	99.69 %	606944	*No OMIM phenotypes*
FAM111B	99.98 %	615584	Poikiloderma, hereditary fibrosing, with tendon contractures, myopathy, and pulmonary fibrosis, 615704 (3), Autosomal dominant
FARSA	100 %	602918	?Rajab interstitial lung disease with brain calcifications 2, 619013 (3), Autosomal recessive
FARSB	99.64 %	609690	Rajab interstitial lung disease with brain calcifications 1, 613658 (3), Autosomal recessive
FBLN5	100 %	604580	Cutis laxa, autosomal recessive, type IA, 219100 (3), Autosomal recessive; Charcot-Marie-Tooth disease, demyelinating, type 1H, 619764 (3), Autosomal dominant; Macular degeneration, age-related, 3, 608895 (3), Autosomal dominant; Neuropathy, hereditary, with or without age-related macular degeneration, 608895 (3), Autosomal dominant; ?Cutis laxa, autosomal dominant 2, 614434 (3), Autosomal dominant
FGF10	99.95 %	602115	LADD syndrome 3, 620193 (3); Aplasia of lacrimal and salivary glands, 180920 (3), Autosomal dominant
FGFR2	99.99 %	176943	Bent bone dysplasia syndrome, 614592 (3), Autosomal dominant; LADD syndrome 1, 149730 (3), Autosomal dominant; Antley-Bixler syndrome without genital anomalies or disordered steroidogenesis, 207410 (3), Autosomal dominant; Scaphocephaly and Axenfeld-Rieger anomaly (3); Jackson-Weiss syndrome, 123150 (3), Autosomal dominant; Gastric cancer, somatic, 613659 (3); Craniofacial-skeletal-dermatologic dysplasia, 101600 (3), Autosomal dominant; Apert syndrome, 101200 (3), Autosomal dominant; Pfeiffer syndrome, 101600 (3), Autosomal dominant; Craniosynostosis, nonspecific (3); ?Scaphocephaly, maxillary retrusion, and impaired intellectual development, 609579 (3); Beare-Stevenson cutis gyrata syndrome, 123790 (3), Autosomal dominant; Crouzon syndrome, 123500 (3), Autosomal dominant; Saethre-Chotzen syndrome, 101400 (3), Autosomal dominant
FLNA	99.99 %	300017	Otopalatodigital syndrome, type II, 304120 (3), X-linked dominant; Intestinal pseudoobstruction, neuronal, 300048 (3), X-linked recessive; Cardiac valvular dysplasia, X-linked, 314400 (3), X-linked; ?FG syndrome 2, 300321 (3), X-linked; Melnick-Needles syndrome, 309350 (3), X-linked dominant; Terminal osseous dysplasia, 300244 (3), X-linked dominant; Congenital short bowel syndrome, 300048 (3), X-linked recessive; Otopalatodigital syndrome, type I, 311300 (3), X-linked dominant; Heterotopia, periventricular, 1, 300049 (3), X-linked dominant; Frontometaphyseal dysplasia 1, 305620 (3), X-linked recessive
FNIP1	99.87 %	610594	Immunodeficiency 93 and hypertrophic cardiomyopathy, 619705 (3), Autosomal recessive

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
FOXF1	99.99 %	601089	Alveolar capillary dysplasia with misalignment of pulmonary veins, 265380 (3), Autosomal dominant
FOXP3	99.93 %	300292	Immunodysregulation, polyendocrinopathy, and enteropathy, X-linked, 304790 (3), X-linked recessive
GARS1	99.93 %	600287	Spinal muscular atrophy, infantile, James type, 619042 (3), Autosomal dominant; Neuronopathy, distal hereditary motor, type VA, 600794 (3), Autosomal dominant; Charcot-Marie-Tooth disease, type 2D, 601472 (3), Autosomal dominant
GAS2L2	100 %	611398	?Ciliary dyskinesia, primary, 41, 618449 (3), Autosomal recessive
GAS8	100 %	605178	Ciliary dyskinesia, primary, 33, 616726 (3), Autosomal recessive
GATA2	99.99 %	137295	{Leukemia, acute myeloid, susceptibility to}, 601626 (3), Somatic mutation, Autosomal dominant; Emberger syndrome, 614038 (3), Autosomal dominant; Immunodeficiency 21, 614172 (3), Autosomal dominant; {Myelodysplastic syndrome, susceptibility to}, 614286 (3)
GBA	96.92 %	606463	{Lewy body dementia, susceptibility to}, 127750 (3), Autosomal dominant; Gaucher disease, type II, 230900 (3), Autosomal recessive; Gaucher disease, type IIIC, 231005 (3), Autosomal recessive; Gaucher disease, type III, 231000 (3), Autosomal recessive; Gaucher disease, type I, 230800 (3), Autosomal recessive; Gaucher disease, perinatal lethal, 608013 (3), Autosomal recessive; {Parkinson disease, late-onset, susceptibility to}, 168600 (3), Multifactorial, Autosomal dominant
GDF2	100 %	605120	Telangiectasia, hereditary hemorrhagic, type 5, 615506 (3), Autosomal dominant
HCK	100 %	142370	Autoinflammation with pulmonary and cutaneous vasculitis, 620296 (3), Autosomal dominant
HLA-DRB1	95.81 %	142857	{Multiple sclerosis, susceptibility to, 1}, 126200 (3), Multifactorial; {Sarcoidosis, susceptibility to, 1}, 181000 (3), Autosomal dominant
HPS1	100 %	604982	Hermansky-Pudlak syndrome 1, 203300 (3), Autosomal recessive
HPS3	99.91 %	606118	Hermansky-Pudlak syndrome 3, 614072 (3), Autosomal recessive
HPS4	99.98 %	606682	Hermansky-Pudlak syndrome 4, 614073 (3), Autosomal recessive
HPS5	99.91 %	607521	Hermansky-Pudlak syndrome 5, 614074 (3), Autosomal recessive
HPS6	100 %	607522	Hermansky-Pudlak syndrome 6, 614075 (3), Autosomal recessive
IARS1	99.89 %	600709	Growth retardation, impaired intellectual development, hypotonia, and hepatopathy, 617093 (3), Autosomal recessive
IKZF1	99.92 %	603023	Immunodeficiency, common variable, 13, 616873 (3), Autosomal dominant
IL6R	92.46 %	147880	[Interleukin 6, serum level of, QTL], 614752 (3); Hyper-IgE recurrent infection syndrome 5, autosomal recessive, 618944 (3), Autosomal recessive; [Interleukin-6 receptor, soluble, serum level of, QTL], 614689 (3)
IL6ST	99.88 %	600694	Stuve-Wiedemann syndrome 2, 619751 (3), Autosomal recessive; Hyper-IgE recurrent infection syndrome 4A, autosomal dominant, 619752 (3), Autosomal dominant; ?Immunodeficiency 94 with autoinflammation and dysmorphic facies, 619750 (3), Autosomal dominant; Hyper-IgE recurrent infection syndrome 4B, autosomal recessive, 618523 (3), Autosomal recessive
IRF2BP2	100 %	615332	?Immunodeficiency, common variable, 14, 617765 (3), Autosomal dominant
ITCH	95.57 %	606409	Autoimmune disease, multisystem, with facial dysmorphism, 613385 (3), Autosomal recessive
ITGA3	99.86 %	605025	Epidermolysis bullosa, junctional 7, with interstitial lung disease and nephrotic syndrome, 614748 (3), Autosomal recessive
KCNA5	100 %	176267	Atrial fibrillation, familial, 7, 612240 (3), Autosomal dominant
KCNK3	100 %	603220	Pulmonary hypertension, primary, 4, 615344 (3), Autosomal dominant

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KDR	99.86 %	191306	{Hemangioma, capillary infantile, susceptibility to}, 602089 (3), Autosomal dominant; Hemangioma, capillary infantile, somatic, 602089 (3)
KIF15	99.93 %	617569	?Braddock-Carey syndrome 2, 619981 (3), Autosomal recessive
KLF2	99.98 %	602016	*No OMIM phenotypes*
LARS1	99.87 %	151350	?Infantile liver failure syndrome 1, 615438 (3), Autosomal recessive
LBX1	99.99 %	604255	?Central hypoventilation syndrome, congenital, 3, 619483 (3), Autosomal recessive
LRBA	99.76 %	606453	Immunodeficiency, common variable, 8, with autoimmunity, 614700 (3), Autosomal recessive
LTBP4	99.99 %	604710	Cutis laxa, autosomal recessive, type IC, 613177 (3), Autosomal recessive
MARS1	99.97 %	156560	Spastic paraplegia 70, autosomal recessive, 620323 (3), Autosomal recessive; Interstitial lung and liver disease, 615486 (3), Autosomal recessive; ?Trichothiodystrophy 9, nonphotosensitive, 619692 (3), Autosomal recessive; Charcot-Marie-Tooth disease, axonal, type 2U, 616280 (3), Autosomal dominant
MCIDAS	100 %	614086	Ciliary dyskinesia, primary, 42, 618695 (3), Autosomal recessive
MOGS	100 %	601336	Congenital disorder of glycosylation, type IIb, 606056 (3), Autosomal recessive
MS4A1	99.6 %	112210	?Immunodeficiency, common variable, 5, 613495 (3), Autosomal recessive
MUC5B	99.94 %	600770	{Pulmonary fibrosis, idiopathic, susceptibility to}, 178500 (3), Autosomal dominant
MVK	99.97 %	251170	Hyper-IgD syndrome, 260920 (3), Autosomal recessive; Porokeratosis 3, multiple types, 175900 (3), Autosomal dominant; Mevalonic aciduria, 610377 (3), Autosomal recessive
MYO1H	99.99 %	614636	?Central hypoventilation syndrome, congenital, 2, and autonomic dysfunction, 619482 (3), Autosomal recessive
NAF1	99.94 %	617868	Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 7, 620365 (3), Autosomal dominant
NEK10	96.92 %	618726	Ciliary dyskinesia, primary, 44, 618781 (3), Autosomal recessive
NF1	99.88 %	613113	Watson syndrome, 193520 (3), Autosomal dominant; Leukemia, juvenile myelomonocytic, 607785 (3), Somatic mutation, Autosomal dominant; Neurofibromatosis, familial spinal, 162210 (3), Autosomal dominant; Neurofibromatosis, type 1, 162200 (3), Autosomal dominant; Neurofibromatosis-Noonan syndrome, 601321 (3), Autosomal dominant
NFKB1	99.8 %	164011	Immunodeficiency, common variable, 12, 616576 (3), Autosomal dominant
NFKB2	99.98 %	164012	Immunodeficiency, common variable, 10, 615577 (3), Autosomal dominant
NHLRC2	99.83 %	618277	FINCA syndrome, 618278 (3), Autosomal recessive
NHP2	99.96 %	606470	Dyskeratosis congenita, autosomal recessive 2, 613987 (3), Autosomal recessive
NKX2-1	100 %	600635	Chorea, hereditary benign, 118700 (3), Autosomal dominant; {Thyroid cancer, nonmedullary, 1}, 188550 (3), Autosomal dominant; Choreoathetosis, hypothyroidism, and neonatal respiratory distress, 610978 (3), Autosomal dominant
NME8	99.83 %	607421	?Ciliary dyskinesia, primary, 6, 610852 (3), Autosomal recessive
NOD2	99.98 %	605956	Blau syndrome, 186580 (3), Autosomal dominant; {Yao syndrome}, 617321 (3), Multifactorial; {Inflammatory bowel disease 1, Crohn disease}, 266600 (3), Multifactorial
NOP10	99.99 %	606471	?Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 9, 620400 (3), Autosomal dominant; ?Cataracts, hearing impairment, nephrotic syndrome, and enterocolitis 2, 620425 (3), Autosomal recessive; ?Dyskeratosis congenita, autosomal recessive 1, 224230 (3), Autosomal recessive
NOTCH3	99.99 %	600276	Lateral meningocele syndrome, 130720 (3), Autosomal dominant; ?Myofibromatosis, infantile 2, 615293 (3), Autosomal dominant; Cerebral arteriopathy with subcortical infarcts and leukoencephalopathy 1, 125310 (3), Autosomal dominant

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NR3C2	100 %	600983	Pseudohypoaldosteronism type I, autosomal dominant, 177735 (3), Autosomal dominant; Hypertension, early-onset, autosomal dominant, with exacerbation in pregnancy, 605115 (3)
NSMCE3	100 %	608243	Lung disease, immunodeficiency, and chromosome breakage syndrome, 617241 (3), Autosomal recessive
OAS1	99.96 %	164350	Immunodeficiency 100 with pulmonary alveolar proteinosis and hypogammaglobulinemia, 618042 (3), Autosomal dominant
PARN	99.75 %	604212	Dyskeratosis congenita, autosomal recessive 6, 616353 (3), Autosomal recessive; Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 4, 616371 (3), Autosomal dominant
PGM3	99.94 %	172100	Immunodeficiency 23, 615816 (3), Autosomal recessive
PHOX2B	99.98 %	603851	{Neuroblastoma, susceptibility to, 2}, 613013 (3); Neuroblastoma with Hirschsprung disease, 613013 (3); Central hypoventilation syndrome, congenital, 1, with or without Hirschsprung disease, 209880 (3), Autosomal dominant
PIK3CD	99.99 %	602839	Immunodeficiency 14A, autosomal dominant, 615513 (3), Autosomal dominant; Immunodeficiency 14B, autosomal recessive, 619281 (3), Autosomal recessive; ?Roifman-Chitayat syndrome, digenic, 613328 (3), Digenic recessive
PIK3CG	99.72 %	601232	Immunodeficiency 97 with autoinflammation, 619802 (3), Autosomal recessive
PIK3R1	99.86 %	171833	Immunodeficiency 36, 616005 (3), Autosomal dominant; ?Agammaglobulinemia 7, autosomal recessive, 615214 (3), Autosomal recessive; SHORT syndrome, 269880 (3), Autosomal dominant
POLD1	99.96 %	174761	Mandibular hypoplasia, deafness, progeroid features, and lipodystrophy syndrome, 615381 (3), Autosomal dominant; {Colorectal cancer, susceptibility to, 10}, 612591 (3), Autosomal dominant
POT1	99.91 %	606478	{Glioma susceptibility 9}, 616568 (3), Autosomal dominant; ?Cerebroretinal microangiopathy with calcifications and cysts 3, 620368 (3), Autosomal recessive; {Melanoma, cutaneous malignant, susceptibility to, 10}, 615848 (3), Autosomal dominant; ?Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 8, 620367 (3), Autosomal dominant
POU2AF1	99.43 %	601206	*No OMIM phenotypes*
PTEN	99.89 %	601728	{Glioma susceptibility 2}, 613028 (3), Autosomal dominant; {Meningioma}, 607174 (3), Autosomal dominant; Cowden syndrome 1, 158350 (3), Autosomal dominant; Lhermitte-Duclos disease, 158350 (3), Autosomal dominant; Prostate cancer, somatic, 176807 (3); Macrocephaly/autism syndrome, 605309 (3), Autosomal dominant
RAC2	99.99 %	602049	Immunodeficiency 73A with defective neutrophil chemotaxix and leukocytosis, 608203 (3), Autosomal dominant; ?Immunodeficiency 73C with defective neutrophil chemotaxis and hypogammaglobulinemia, 618987 (3), Autosomal recessive; Immunodeficiency 73B with defective neutrophil chemotaxis and lymphopenia, 618986 (3), Autosomal dominant
RASA1	99.05 %	139150	Capillary malformation-arteriovenous malformation 1, 608354 (3), Autosomal dominant; Basal cell carcinoma, somatic, 605462 (3)
RFX5	99.88 %	601863	Bare lymphocyte syndrome, type II, complementation group C, 209920 (3), Autosomal recessive; Bare lymphocyte syndrome, type II, complementation group E, 209920 (3), Autosomal recessive
RFXANK	100 %	603200	Bare lymphocyte syndrome, type II, complementation group B, 209920 (3), Autosomal recessive
RFXAP	99.98 %	601861	Bare lymphocyte syndrome, type II, complementation group D, 209920 (3), Autosomal recessive
RGPD4	71.96 %	612707	*No OMIM phenotypes*

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RNF168	99.97 %	612688	RIDDLE syndrome, 611943 (3), Autosomal recessive
RSPH3	99.94 %	615876	Ciliary dyskinesia, primary, 32, 616481 (3), Autosomal recessive
RSPH4A	99.95 %	612647	Ciliary dyskinesia, primary, 11, 612649 (3), Autosomal recessive
RSPH9	99.99 %	612648	Ciliary dyskinesia, primary, 12, 612650 (3), Autosomal recessive
RTEL1	100 %	608833	Dyskeratosis congenita, autosomal dominant 4, 615190 (3), Autosomal recessive, Autosomal dominant; Dyskeratosis congenita, autosomal recessive 5, 615190 (3), Autosomal recessive, Autosomal dominant; Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 3, 616373 (3), Autosomal dominant
SCNN1A	100 %	600228	Pseudohypoaldosteronism, type IB1, autosomal recessive, 264350 (3), Autosomal recessive; ?Liddle syndrome 3, 618126 (3), Autosomal dominant; Bronchiectasis with or without elevated sweat chloride 2, 613021 (3), Autosomal dominant
SCNN1B	99.38 %	600760	Bronchiectasis with or without elevated sweat chloride 1, 211400 (3), Autosomal dominant; Pseudohypoaldosteronism, type IB2, autosomal recessive, 620125 (3), Autosomal recessive; Liddle syndrome 1, 177200 (3), Autosomal dominant
SCNN1G	99.94 %	600761	Bronchiectasis with or without elevated sweat chloride 3, 613071 (3), Autosomal dominant; Pseudohypoaldosteronism, type IB3, autosomal recessive, 620126 (3), Autosomal recessive; Liddle syndrome 2, 618114 (3), Autosomal dominant
SEC61A1	99.99 %	609213	Tubulointerstitial kidney disease, autosomal dominant, 5, 617056 (3), Autosomal dominant
SERPINA1	100 %	107400	Hemorrhagic diathesis due to antithrombin Pittsburgh, 613490 (3), Autosomal recessive; Emphysema due to AAT deficiency, 613490 (3), Autosomal recessive; Emphysema-cirrhosis, due to AAT deficiency, 613490 (3), Autosomal recessive
SFTPA1	99.99 %	178630	Interstitial lung disease 1, 619611 (3), Autosomal recessive, Autosomal dominant
SFTPA2	99.81 %	178642	Interstitial lung disease 2, 178500 (3), Autosomal dominant
SFTPB	99.99 %	178640	Surfactant metabolism dysfunction, pulmonary, 1, 265120 (3), Autosomal recessive
SFTPC	99.99 %	178620	Surfactant metabolism dysfunction, pulmonary, 2, 610913 (3), Autosomal dominant
SFTPD	99.62 %	178635	*No OMIM phenotypes*
SH3KBP1	99.95 %	300374	?Immunodeficiency 61, 300310 (3), X-linked recessive
SLC34A2	99.99 %	604217	Pulmonary alveolar microlithiasis, 265100 (3), Autosomal recessive
SLC7A7	99.99 %	603593	Lysinuric protein intolerance, 222700 (3), Autosomal recessive
SMAD4	99.97 %	600993	Pancreatic cancer, somatic, 260350 (3); Myhre syndrome, 139210 (3), Autosomal dominant; Polyposis, juvenile intestinal, 174900 (3), Autosomal dominant; Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome, 175050 (3), Autosomal dominant
SMAD9	99.99 %	603295	Pulmonary hypertension, primary, 2, 615342 (3), Autosomal dominant
SMPD1	100 %	607608	Niemann-Pick disease, type B, 607616 (3), Autosomal recessive; Niemann-Pick disease, type A, 257200 (3), Autosomal recessive
SOX17	100 %	610928	Vesicoureteral reflux 3, 613674 (3), Autosomal dominant
SPDL1	99.92 %	616401	*No OMIM phenotypes*
SPINK5	99.91 %	605010	Netherton syndrome, 256500 (3), Autosomal recessive
STAT3	99.97 %	102582	Hyper-IgE recurrent infection syndrome, 147060 (3), Autosomal dominant; Autoimmune disease, multisystem, infantile-onset, 1, 615952 (3), Autosomal dominant
STAT5B	99.5 %	604260	Growth hormone insensitivity with immune dysregulation 1, autosomal recessive, 245590 (3), Autosomal recessive; Growth hormone insensitivity with immune dysregulation 2, autosomal dominant, 618985 (3), Autosomal dominant; Leukemia, acute promyelocytic, somatic, 102578 (3)
STING1	99.87 %	612374	STING-associated vasculopathy, infantile-onset, 615934 (3), Autosomal dominant

Gene	% at least 20 x covered*	OMIM gene id	OMIM Phenotypes
STK36	99.98 %	607652	?Ciliary dyskinesia, primary, 46, 619436 (3), Autosomal recessive
TBX4	99.96 %	601719	Ischiocoxopodopatellar syndrome with or without pulmonary arterial hypertension, 147891 (3), Autosomal dominant; Amelia, posterior, with pelvic and pulmonary hypoplasia syndrome, 601360 (3), Autosomal recessive
TERC	98.59 %	602322	Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 2, 614743 (3), Autosomal dominant; Dyskeratosis congenita, autosomal dominant 1, 127550 (3), Autosomal dominant
TERT	100 %	187270	Dyskeratosis congenita, autosomal dominant 2, 613989 (3), Autosomal recessive, Autosomal dominant; Dyskeratosis congenita, autosomal recessive 4, 613989 (3), Autosomal recessive, Autosomal dominant; Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 1, 614742 (3), Autosomal dominant; {Melanoma, cutaneous malignant, 9}, 615134 (3), Autosomal dominant; {Leukemia, acute myeloid}, 601626 (3), Somatic mutation, Autosomal dominant
TGFBR1	99.94 %	190181	{Multiple self-healing squamous epithelioma, susceptibility to}, 132800 (3), Autosomal dominant; Loeys-Dietz syndrome 1, 609192 (3), Autosomal dominant
TGFBR2	99.98 %	190182	Loeys-Dietz syndrome 2, 610168 (3), Autosomal dominant; Colorectal cancer, hereditary nonpolyposis, type 6, 614331 (3); Esophageal cancer, somatic, 133239 (3)
TINF2	100 %	604319	Dyskeratosis congenita, autosomal dominant 3, 613990 (3), Autosomal dominant; Revesz syndrome, 268130 (3), Autosomal dominant
TNFRSF13B	99.43 %	604907	Immunodeficiency, common variable, 2, 240500 (3), Autosomal recessive, Autosomal dominant; Immunoglobulin A deficiency 2, 609529 (3)
TNFRSF13C	99.99 %	606269	Immunodeficiency, common variable, 4, 613494 (3), Autosomal recessive
TNFSF12	100 %	602695	*No OMIM phenotypes*
TNFSF13	100 %	604472	*No OMIM phenotypes*
TRNT1	99.97 %	612907	Sideroblastic anemia with B-cell immunodeficiency, periodic fevers, and developmental delay, 616084 (3), Autosomal recessive; Retinitis pigmentosa and erythrocytic microcytosis, 616959 (3), Autosomal recessive
TSC1	99.99 %	605284	Focal cortical dysplasia, type II, somatic, 607341 (3); Tuberous sclerosis-1, 191100 (3), Autosomal dominant; Lymphangioleiomyomatosis, 606690 (3)
TSC2	99.98 %	191092	Lymphangioleiomyomatosis, somatic, 606690 (3); ?Focal cortical dysplasia, type II, somatic, 607341 (3); Tuberous sclerosis-2, 613254 (3), Autosomal dominant
WRAP53	100 %	612661	Dyskeratosis congenita, autosomal recessive 3, 613988 (3), Autosomal recessive
YARS1	99.29 %	603623	Infantile-onset multisystem neurologic, endocrine, and pancreatic disease 2, 619418 (3), Autosomal recessive; Charcot-Marie-Tooth disease, dominant intermediate C, 608323 (3), Autosomal dominant
ZCCHC8	99.95 %	616381	?Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 5, 618674 (3), Autosomal dominant
ZNF341	100 %	618269	Hyper-IgE recurrent infection syndrome 3, autosomal recessive, 618282 (3), Autosomal recessive
ZNFX1	99.99 %	618931	Immunodeficiency 91 and hyperinflammation, 619644 (3), Autosomal recessive

Explanation

OMIM release used for OMIM disease identifiers and descriptions: **2023-07-31**

Gene symbols used are according to the HGNC guidelines (corresponding to Ensembl database release 105).

Each Phenotype is followed by its MIM number, phenotype mapping key and inheritance pattern.

Possible phenotype mapping keys

(1) the disorder is placed on the map based on its association with a gene, but the underlying defect is not known

(2) the disorder has been placed on the map by linkage; no mutation has been found

(3) the molecular basis for the disorder is known; a mutation has been found in the gene

(4) a contiguous gene deletion or duplication syndrome, multiple genes are deleted or duplicated causing the phenotype

Brackets, "[ ]", indicate "nondiseases," mainly genetic variations that lead to apparently abnormal laboratory test values (e.g., dysalbuminemic euthyroidal hyperthyroxinemia).

Braces, "{ }", indicate mutations that contribute to susceptibility to multifactorial disorders (e.g., diabetes, asthma) or to susceptibility to infection (e.g., malaria).

A question mark, "?", before the phenotype name indicates that the relationship between the phenotype and gene is provisional. More details about this relationship are provided in the comment field of the map and in the gene and phenotype OMIM entries.

* The column '% at least 20 x covered' shows the percentage of the coding sequence (+/-20 nucleotides of the flanking introns) of that gene that is on average at least 20 x covered. This according to the experience with exome sequencing in our laboratory and based on the current method.