Respiratory Disorders panel

versie	v2 (137 genen)	Centrum voor Medische Genetica Gent

Gene	OMIM gene ID	Associated phenotype, OMIM phenotype ID, phenotype mapping key and inheritance pattern
ABCA3	601615	Surfactant metabolism dysfunction, pulmonary, 3, 610921 (3), Autosomal recessive
ABCC8	600509	Diabetes mellitus, permanent neonatal 3, with or without neurologic features, 618857 (3), Autosomal dominant, Autosomal recessive; 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 dominant, Autosomal recessive
ACVRL1	601284	Telangiectasia, hereditary hemorrhagic, type 2, 600376 (3), Autosomal dominant
AP3B1	603401	Hermansky-Pudlak syndrome 2, 608233 (3), Autosomal recessive
AQP1	107776	[Aquaporin-1 deficiency], 110450 (3); [Blood group, Colton], 110450 (3)
ARHGEF1	601855	?Immunodeficiency 62, 618459 (3), Autosomal recessive
ASAH1	613468	Spinal muscular atrophy with progressive myoclonic epilepsy, 159950 (3), Autosomal recessive; Farber lipogranulomatosis, 228000 (3), Autosomal recessive
ASCL1	100790	Haddad syndrome, 209880 (3), Autosomal dominant; Central hypoventilation syndrome, congenital, 209880 (3), Autosomal dominant
ATP13A3	610232	Pulmonary hypertension, primary, 5, 265400 (3), Autosomal recessive

ATP6AP1	300197	Immunodeficiency 47, 300972 (3), X-linked recessive
BDNF	113505	No OMIM phenotype
BLOC1S3	609762	Hermansky-Pudlak syndrome 8, 614077 (3), Autosomal recessive
BLOC1S6	604310	?Hermansky-Pudlak syndrome 9, 614171 (3), Autosomal recessive
BMP10	608748	No OMIM phenotype
BMPR1B	603248	Acromesomelic dysplasia 3, 609441 (3), Autosomal recessive; Brachydactyly, type A2, 112600 (3), Autosomal dominant; Brachydactyly, type A1, D, 616849 (3), Autosomal dominant
BMPR2	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	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
CAV1	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	613798	Ciliary dyskinesia, primary, 14, 613807 (3), Autosomal recessive
CCDC40	613799	Ciliary dyskinesia, primary, 15, 613808 (3), Autosomal recessive
CD19	107265	Immunodeficiency, common variable, 3, 613493 (3), Autosomal recessive
CD81	186845	Immunodeficiency, common variable, 6, 613496 (3), Autosomal recessive

CIITA	600005	{Rheumatoid arthritis, susceptibility to}, 180300 (3); Bare lymphocyte syndrome, type II, complementation group A, 209920 (3), Autosomal recessive
CLEC1A	606782	{Aspergillosis, susceptibility to}, 614079 (3)
CLEC7A	606264	Candidiasis, familial, 4, autosomal recessive, 613108 (3), Autosomal recessive; {Aspergillosis, susceptibility to}, 614079 (3)
COPA	601924	{Autoimmune interstitial lung, joint, and kidney disease}, 616414 (3), Autosomal dominant
CR2	120650	{Systemic lupus erythematosus, susceptibility to, 9}, 610927 (3); Immunodeficiency, common variable, 7, 614699 (3), Autosomal recessive
CSF2RA	306250	Surfactant metabolism dysfunction, pulmonary, 4, 300770 (3)
CSF2RB	138981	Surfactant metabolism dysfunction, pulmonary, 5, 614370 (3), Autosomal recessive
CTLA4	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	611537	?Immunodeficiency 99 with hypogammaglobulinemia and autoimmune cytopenias, 619846 (3), Autosomal recessive
DKC1	300126	Dyskeratosis congenita, X-linked, 305000 (3), X-linked recessive
DNAAF1	613190	Ciliary dyskinesia, primary, 13, 613193 (3), Autosomal recessive
DNAAF2	612517	Ciliary dyskinesia, primary, 10, 612518 (3), Autosomal recessive
DNAH11	603339	Ciliary dyskinesia, primary, 7, with or without situs inversus, 611884 (3), Autosomal recessive

DNAH5	603335	Ciliary dyskinesia, primary, 3, with or without situs inversus, 608644 (3), Autosomal recessive
DNAI1	604366	Ciliary dyskinesia, primary, 1, with or without situs inversus, 244400 (3), Autosomal recessive
DNAI2	605483	Ciliary dyskinesia, primary, 9, with or without situs inversus, 612444 (3), Autosomal recessive
DNAL1	610062	Ciliary dyskinesia, primary, 16, 614017 (3), Autosomal recessive
DOCK8	611432	Hyper-IgE recurrent infection syndrome, autosomal recessive, 243700 (3), Autosomal recessive
DTNBP1	607145	Hermansky-Pudlak syndrome 7, 614076 (3), Autosomal recessive
EFEMP2	604633	Cutis laxa, autosomal recessive, type IB, 614437 (3), Autosomal recessive
EIF2AK4	609280	Pulmonary venoocclusive disease 2, 234810 (3), Autosomal recessive
ELMOD2	610196	No OMIM phenotype
ELN	130160	Cutis laxa, autosomal dominant, 123700 (3), Autosomal dominant; Supravalvar aortic stenosis, 185500 (3), Autosomal dominant
ENG	131195	Telangiectasia, hereditary hemorrhagic, type 1, 187300 (3), Autosomal dominant
ERBIN	606944	No OMIM phenotype
FAM111B	615584	Poikiloderma, hereditary fibrosing, with tendon contractures, myopathy, and pulmonary fibrosis, 615704 (3), Autosomal dominant
FARSA	602918	?Rajab interstitial lung disease with brain calcifications 2, 619013 (3), Autosomal recessive
FARSB	609690	Rajab interstitial lung disease with brain calcifications 1, 613658 (3), Autosomal recessive

FBLN5	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
FLNA	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	610594	Immunodeficiency 93 and hypertrophic cardiomyopathy, 619705 (3), Autosomal recessive
FOXF1	601089	Alveolar capillary dysplasia with misalignment of pulmonary veins, 265380 (3), Autosomal dominant
FOXP3	300292	Immunodysregulation, polyendocrinopathy, and enteropathy, X-linked, 304790 (3), X-linked recessive
GARS1	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
GATA2	137295	{Leukemia, acute myeloid, susceptibility to}, 601626 (3), Autosomal dominant, Somatic mutation; Emberger syndrome, 614038 (3), Autosomal dominant; Immunodeficiency 21, 614172 (3), Autosomal dominant; {Myelodysplastic syndrome, susceptibility to}, 614286 (3)

GBA1 (GBA)	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	605120	Telangiectasia, hereditary hemorrhagic, type 5, 615506 (3), Autosomal dominant
HPS1	604982	Hermansky-Pudlak syndrome 1, 203300 (3), Autosomal recessive
HPS3	606118	Hermansky-Pudlak syndrome 3, 614072 (3), Autosomal recessive
HPS4	606682	Hermansky-Pudlak syndrome 4, 614073 (3), Autosomal recessive
HPS5	607521	Hermansky-Pudlak syndrome 5, 614074 (3), Autosomal recessive
HPS6	607522	Hermansky-Pudlak syndrome 6, 614075 (3), Autosomal recessive
IKZF1	603023	Immunodeficiency, common variable, 13, 616873 (3), Autosomal dominant
IL6R	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	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	615332	?Immunodeficiency, common variable, 14, 617765 (3), Autosomal dominant

ITCH	606409	Autoimmune disease, multisystem, with facial dysmorphism, 613385 (3), Autosomal recessive
ITGA3	605025	Epidermolysis bullosa, junctional 7, with interstitial lung disease and nephrotic syndrome, 614748 (3), Autosomal recessive
KCNA5	176267	Atrial fibrillation, familial, 7, 612240 (3), Autosomal dominant
KCNK3	603220	Pulmonary hypertension, primary, 4, 615344 (3), Autosomal dominant
KDR	191306	{Hemangioma, capillary infantile, susceptibility to}, 602089 (3), Autosomal dominant; Hemangioma, capillary infantile, somatic, 602089 (3)
LRBA	606453	Immunodeficiency, common variable, 8, with autoimmunity, 614700 (3), Autosomal recessive
LTBP4	604710	Cutis laxa, autosomal recessive, type IC, 613177 (3), Autosomal recessive
MARS1	156560	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
MOGS	601336	Congenital disorder of glycosylation, type IIb, 606056 (3), Autosomal recessive
MS4A1	112210	?Immunodeficiency, common variable, 5, 613495 (3), Autosomal recessive
MUC5B	600770	{Pulmonary fibrosis, idiopathic, susceptibility to}, 178500 (3), Autosomal dominant
NFKB1	164011	Immunodeficiency, common variable, 12, 616576 (3), Autosomal dominant
NFKB2	164012	Immunodeficiency, common variable, 10, 615577 (3), Autosomal dominant
NKX2-1	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	607421	Ciliary dyskinesia, primary, 6, 610852 (3), Autosomal recessive
NOD2	605956	Blau syndrome, 186580 (3), Autosomal dominant; {Yao syndrome}, 617321 (3), Multifactorial; {Inflammatory bowel disease 1, Crohn disease}, 266600 (3), Multifactorial
NOTCH3	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
NSMCE3	608243	Lung disease, immunodeficiency, and chromosome breakage syndrome, 617241 (3), Autosomal recessive
OAS1	164350	Immunodeficiency 100 with pulmonary alveolar proteinosis and hypogammaglobulinemia, 618042 (3), Autosomal dominant
PARN	604212	Dyskeratosis congenita, autosomal recessive 6, 616353 (3), Autosomal recessive; Pulmonary fibrosis and/or bone marrow failure, telomere-related, 4, 616371 (3), Autosomal dominant
PGM3	172100	Immunodeficiency 23, 615816 (3), Autosomal recessive
PIK3CD	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	601232	Immunodeficiency 97 with autoinflammation, 619802 (3), Autosomal recessive
PIK3R1	171833	Immunodeficiency 36, 616005 (3), Autosomal dominant; ?Agammaglobulinemia 7, autosomal recessive, 615214 (3), Autosomal recessive; SHORT syndrome, 269880 (3), Autosomal dominant
POU2AF1	601206	No OMIM phenotype

PTEN	601728	{Glioma susceptibility 2}, 613028 (3); {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	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
RFX5	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	603200	MHC class II deficiency, complementation group B, 209920 (3), Autosomal recessive
RFXAP	601861	Bare lymphocyte syndrome, type II, complementation group D, 209920 (3), Autosomal recessive
RGPD4	612707	No OMIM phenotype
RSPH4A	612647	Ciliary dyskinesia, primary, 11, 612649 (3)
RSPH9	612648	Ciliary dyskinesia, primary, 12, 612650 (3)
RTEL1	608833	Pulmonary fibrosis and/or bone marrow failure, telomere-related, 3, 616373 (3), Autosomal dominant; Dyskeratosis congenita, autosomal dominant 4, 615190 (3), Autosomal dominant, Autosomal recessive; Dyskeratosis congenita, autosomal recessive 5, 615190 (3), Autosomal dominant, Autosomal recessive
SCNN1A	600228	Pseudohypoaldosteronism, type I, 264350 (3), Autosomal recessive; ?Liddle syndrome 3, 618126 (3), Autosomal dominant; Bronchiectasis with or without elevated sweat chloride 2, 613021 (3), Autosomal dominant

SCNN1B	600760	Bronchiectasis with or without elevated sweat chloride 1, 211400 (3), Autosomal dominant; Pseudohypoaldosteronism, type I, 264350 (3), Autosomal recessive; Liddle syndrome 1, 177200 (3), Autosomal dominant
SCNN1G	600761	Bronchiectasis with or without elevated sweat chloride 3, 613071 (3), Autosomal dominant; Pseudohypoaldosteronism, type I, 264350 (3), Autosomal recessive; Liddle syndrome 2, 618114 (3), Autosomal dominant
SEC61A1	609213	Tubulointerstitial kidney disease, autosomal dominant, 5, 617056 (3), Autosomal dominant
SERPINA1	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	178630	Interstitial lung disease 1, 619611 (3), Autosomal dominant, Autosomal recessive
SFTPA2	178642	Interstitial lung disease 2, 178500 (3), Autosomal dominant
SFTPB	178640	Surfactant metabolism dysfunction, pulmonary, 1, 265120 (3), Autosomal recessive
SFTPC	178620	Surfactant metabolism dysfunction, pulmonary, 2, 610913 (3), Autosomal dominant
SFTPD	178635	No OMIM phenotype
SH3KBP1	300374	?Immunodeficiency 61, 300310 (3), X-linked recessive
SLC34A2	604217	Pulmonary alveolar microlithiasis, 265100 (3), Autosomal recessive
SLC7A7	603593	Lysinuric protein intolerance, 222700 (3), Autosomal recessive
SMAD4	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	603295	Pulmonary hypertension, primary, 2, 615342 (3), Autosomal dominant
SMPD1	607608	Niemann-Pick disease, type B, 607616 (3), Autosomal recessive; Niemann-Pick disease, type A, 257200 (3), Autosomal recessive
SOX17	610928	Vesicoureteral reflux 3, 613674 (3), Autosomal dominant
SPINK5	605010	Netherton syndrome, 256500 (3), Autosomal recessive
STAT3	102582	Hyper-IgE recurrent infection syndrome, 147060 (3), Autosomal dominant; Autoimmune disease, multisystem, infantile-onset, 1, 615952 (3), Autosomal dominant
STAT5B	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	612374	STING-associated vasculopathy, infantile-onset, 615934 (3), Autosomal dominant
TBX4	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	602322	{Pulmonary fibrosis, idiopathic, susceptibility to}, 614743 (3), Autosomal dominant; Dyskeratosis congenita, autosomal dominant 1, 127550 (3), Autosomal dominant; {Aplastic anemia}, 614743 (3), Autosomal dominant
TERT	187270	Dyskeratosis congenita, autosomal dominant 2, 613989 (3), Autosomal dominant, Autosomal recessive; Dyskeratosis congenita, autosomal recessive 4, 613989 (3), Autosomal dominant, Autosomal recessive; {Melanoma, cutaneous malignant, 9}, 615134 (3); Pulmonary fibrosis and/or bone marrow failure, telomere-related, 1, 614742 (3), Autosomal dominant; {Leukemia, acute myeloid}, 601626 (3), Autosomal dominant, Somatic mutation

TGFBR1	190181	{Multiple self-healing squamous epithelioma, susceptibility to}, 132800 (3), Autosomal dominant; Loeys-Dietz syndrome 1, 609192 (3), Autosomal dominant
TGFBR2	190182	Loeys-Dietz syndrome 2, 610168 (3), Autosomal dominant; Colorectal cancer, hereditary nonpolyposis, type 6, 614331 (3); Esophageal cancer, somatic, 133239 (3)
TINF2	604319	Dyskeratosis congenita, autosomal dominant 3, 613990 (3), Autosomal dominant; Revesz syndrome, 268130 (3), Autosomal dominant
TNFRSF13B	604907	Immunodeficiency, common variable, 2, 240500 (3), Autosomal dominant, Autosomal recessive; Immunoglobulin A deficiency 2, 609529 (3)
TNFRSF13C	606269	Immunodeficiency, common variable, 4, 613494 (3), Autosomal recessive
TNFSF12	602695	No OMIM phenotype
TNFSF13	604472	No OMIM phenotype
TRNT1	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	605284	Focal cortical dysplasia, type II, somatic, 607341 (3); Tuberous sclerosis-1, 191100 (3), Autosomal dominant; Lymphangioleiomyomatosis, 606690 (3)
TSC2	191092	Lymphangioleiomyomatosis, somatic, 606690 (3); ?Focal cortical dysplasia, type II, somatic, 607341 (3); Tuberous sclerosis-2, 613254 (3), Autosomal dominant
ZNF341	618269	Hyper-IgE recurrent infection syndrome 3, autosomal recessive, 618282 (3), Autosomal recessive

Gene symbols used are according to the HGNC guidelines. For some genes a previously HGNC-approved symbol is in brackets.
Each phenotype is followed by its MIM number, phenotype mapping key and inheritance pattern.
OMIM release used for OMIM disease identifiers and descriptions: August 24, 2022

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.