

Genetics of chronic obstructive pulmonary disease: a succinct review, future avenues and prospective clinical applications
Supplementary material



Supplementary Figure 1. Genetic loci identified by genome-wide linkage studies. The loci are colored based on the population where they were found.

Supplementary Table 1. List of genes included in FIGURE 1.

Gene	Full names	Chromosome	Ref.
<i>A2M</i>	α 2-macroglobulin	12	[1]
<i>ACE</i>	Angiotensin I converting enzyme (peptidyl-dipeptidase A) 1	17	[2]
<i>ADAM33</i>	ADAM metallopeptidase domain 33	20	[3–5]
<i>ADRB2</i>	Adrenergic β -2-receptor, surface	5	[6–10]
<i>CD63</i>	CD63 molecule	12	[11]
<i>CDKN1A (p21)</i>	Cyclin-dependent kinase inhibitor 1A (p21, Cip1)	6	[12]
<i>CFTR</i>	Cystic fibrosis transmembrane conductance regulator (ATP-binding cassette sub-family C, member 7)	7	[10,13–20]
<i>CSF2</i>	Colony stimulating factor 2 (granulocyte-macrophage)	5	[21,22]
<i>CSF3</i>	Colony stimulating factor 3 (granulocyte)	17	[21]
<i>CYP1A1</i>	Cytochrome P450, family 1, subfamily A, polypeptide 1	15	[23–25]
<i>CYP1A2</i>	Cytochrome P450, family 1, subfamily A, polypeptide 2	15	[24]
<i>CYP2E1</i>	Cytochrome P450, family 2, subfamily E, polypeptide 1	10	[25,26]
<i>DEFB1</i>	Defensin β 1	8	[27,28]
<i>ELN</i>	Elastin (supravalvular aortic stenosis, Williams-Beuren syndrome)	7	[29–31]
<i>EPHX1</i>	Epoxide hydrolase 1, microsomal (xenobiotic)	1	[6,10,25,29,30,32–47]
<i>GC</i>	Group-specific component (vitamin D binding protein)	4	[10,29,30,35,40,48–55]
<i>GSTM1</i>	Glutathione S-transferase M1	1	[25,29,30,32,35,45,56–64]
<i>GSTO2</i>	Glutathione S-transferase omega 2	10	[10]
<i>GSTP1</i>	Glutathione S-transferase pi 1	11	[10,25,29,30,32,33,35,36,42,44,57–59,61–63,65–68]
<i>GSTT1</i>	Glutathione S-transferase θ 1	22	[25,32,45,57–59,61–63,69]
<i>HCK</i>	Hemopoietic cell kinase	20	[70]
<i>HLA</i>	Classical class 11 subregion of the MHC	6	[51,71,72]
<i>HMOX1</i>	Heme oxygenase (decycling) 1	22	[10,30,35,62,73–76]
<i>IFNG</i>	Interferon- γ	12	[77]
<i>IL10</i>	Interleukin-10	1	[10,22,33,78–83]
<i>IL13</i>	Interleukin-13	5	[7,22,84–89]
<i>IL1B</i>	Interleukin-1 β	2	[22,79,82,90–94]
<i>IL1RN</i>	Interleukin-1 receptor antagonist	2	[79,82,91–94]
<i>IL4</i>	Interleukin-4	5	[7,22]
<i>IL4R</i>	Interleukin-4 receptor	16	[85]
<i>IL6</i>	Interleukin-6	7	[79,83,92]
<i>IL8</i>	Interleukin-8	4	[22,79,82,95,96]
<i>IL8RA</i>	Interleukin-8 receptor α	2	[22,29,30,97]
<i>IL8RB (CXCR2)</i>	Interleukin-8 receptor β	2	[22,29,30,96]
<i>LTA</i>	Lymphotoxin α (TNF superfamily, member 1)	6	[22,79,83,98–103]
<i>LTBP4</i>	Latent transforming growth factor β binding protein 4	19	[29,30]
<i>MMP1</i>	Matrix metallopeptidase 1 (interstitial collagenase)	11	[10,29,30,35,104,105]
<i>MMP12</i>	Matrix metallopeptidase 12 (macrophage elastase)	11	[29,30,104]
<i>MMP14</i>	Matrix metallopeptidase 14 (membrane-inserted)	14	[106]
<i>MMP9</i>	Matrix metallopeptidase 9 (gelatinase B, 92 kDa gelatinase, 92 kDa type IV collagenase)	20	[10,30,35,104,105,107–109]
<i>MSR1</i>	Macrophage scavenger receptor 1	8	[27]
<i>NAT2</i>	<i>N</i> -acetyltransferase 2 (arylamine <i>N</i> -acetyltransferase)	8	[26]
<i>NOS3</i>	Nitric oxide synthase 3 (endothelial cell)	7	[33,110,111]
<i>OR4X1</i>	Olfactory receptor, family 4, subfamily X, member 1	11	[112]
<i>PDE4D</i>	Phosphodiesterase 4D, cAMP-specific (phosphodiesterase E3 dunce homolog, <i>Drosophila</i>)	5	[113]
<i>SERPINA1</i>	Serpin peptidase inhibitor, clade A (α -1 antiproteinase, antitrypsin), member 1	14	See text

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Gene	Full names	Chromosome	Ref.
<i>SERPINA3</i>	Serpin peptidase inhibitor, clade A (α -1 antiproteinase, antitrypsin), member 3	14	[10,29,30,33,35,93,114–120]
<i>SERPINE2</i>	Serpin peptidase inhibitor, clade E (nexin, plasminogen activator inhibitor type 1), member 2	2	[10,29,30,33,36,121–124]
<i>SFTPA1</i>	Surfactant, pulmonary-associated protein A1	10	[125]
<i>SFTPB</i>	Surfactant, pulmonary-associated protein B	2	[10,29,30,33,35,36,125–127]
<i>SFTPC</i>	Surfactant, pulmonary-associated protein C	8	[64]
<i>SMOC2</i>	SPARC related modular calcium binding 2	6	[128]
<i>SOD3</i>	Superoxide dismutase 3, extracellular	4	[10,129,130]
<i>TGFB1</i>	Transforming growth factor β 1	19	[10,29,30,33,36,131–136]
<i>TIMP2</i>	TIMP metalloproteinase inhibitor 2	17	[10,29,30,35,137,138]
<i>TNF</i>	Tumor necrosis factor (TNF superfamily, member 2)	6	[6,22,29,30,33,35,40,79,82,83,87,91–93,96,98–103,139–147]
<i>TP53 (p53)</i>	Tumor protein p53	17	[12]

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