

Additional file 1. Interactome of 109 proteins involved in the patient's phenotype and genotype and associated functions, informed in the String Consortium program.

NOS2	Nitric-oxide synthase, inducible; Nitric oxide synthase, inducible; Produces nitric oxide (NO), a messenger molecule with diverse functions throughout the body. In macrophages, NO mediates tumoricidal and bactericidal actions. It also has nitrosylase activity and mediates cysteine S-nitrosylation of cytoplasmic target proteins such PTGS2/COX2 (By similarity). As a component of the iNOS-S100A8/9 transnitrosylase complex involved in the selective inflammatory stimulus-dependent S-nitrosylation of GAPDH on 'Cys-247' implicated in regulation of the GAIT complex activity and probably mul [...] (1153 aa)
PRR23D1	Proline-rich protein 23D1; Proline rich 23 domain containing 1; Belongs to the PRR23 family (279 aa)
SPAG11B	Sperm-associated antigen 11A; Sperm associated antigen 11B; Belongs to the SPAG11 family (133 aa)
C20orf197	Uncharacterized protein C20orf197; Chromosome 20 open reading frame 197 (126 aa)
CDH1	Cadherin-1; Cadherins are calcium-dependent cell adhesion proteins. They preferentially interact with themselves in a homophilic manner in connecting cells; cadherins may thus contribute to sorting heterogeneous cell types. CDH1 is involved in mechanisms regulating cell-cell adhesions, mobility and proliferation of epithelial cells. Has a potent invasive suppressor role. It is a ligand for integrin alpha-E/beta-7 (882 aa)
DNAH6	Dynein heavy chain 6, axonemal; Force generating protein of respiratory cilia. Produces force towards the minus ends of microtubules. Dynein has ATPase activity; the force-producing power stroke is thought to occur on release of ADP (By similarity); it Belongs to the dynein heavy chain family (4158 aa)
TTC8	Tetratricopeptide repeat protein 8; The BBSome complex is thought to function as a coat complex required for sorting of specific membrane proteins to the primary cilia. The BBSome complex is required for ciliogenesis but is dispensable for centriolar satellite function. This ciliogenic function is partly mediated by the Rab8 GDP/GTP exchange factor, which localizes to the basal body and contacts the BBSome. Rab8(GTP) enters the primary cilium and promotes extension of the ciliary membrane. Firstly the BBSome associates with the ciliary membrane and binds to RAB3IP/Rabin8, the guanosyl [...] (531 aa)
DNAH9	Dynein heavy chain 9, axonemal; Force generating protein of respiratory cilia. Produces force towards the minus ends of microtubules. Dynein has ATPase activity; the force-producing power stroke is thought to occur on release of ADP; Dyneins, axonemal (4486 aa)
DHH	Desert hedgehog protein; Intercellular signal essential for a variety of patterning events during development. May function as a spermatocyte survival factor in the testes. Essential for testes development; Hedgehog signaling molecule family (396 aa)
NR5A1	Steroidogenic factor 1; Transcriptional activator. Essential for sexual differentiation and formation of the primary steroidogenic tissues. Binds to the Ad4 site found in the promoter region of steroidogenic P450 genes such as CYP11A, CYP11B and CYP21B. Also regulates the AMH/Muellerian inhibiting substance gene as well as the AHCH and STAR genes. 5'-YCAAGGYC-3' and 5'- RRAGGTCA-3' are the consensus sequences for the recognition by NR5A1. The SFPQ-NONO-NR5A1 complex binds to the CYP17 promoter and regulates basal and cAMP-dependent transcriptional activity. Binds phosphatidylcholine (B [...]) (461 aa)
CCL3	C-C motif chemokine 3; Monokine with inflammatory and chemokinetic properties. Binds to CCR1, CCR4 and CCR5. One of the major HIV-suppressive factors produced by CD8+ T-cells. Recombinant MIP-1-alpha induces a dose-dependent inhibition of different strains of HIV-1, HIV-2, and simian immunodeficiency virus (SIV); Belongs to the intercrine beta (chemokine CC) family (92 aa)
GTF2H2C	General transcription factor IIH subunit 2-like protein; Component of the core-TFIID basal transcription factor involved in nucleotide excision repair (NER) of DNA and, when complexed to CAK, in RNA transcription by RNA polymerase II (395 aa)
PRODH	Proline dehydrogenase 1, mitochondrial; Converts proline to delta-1-pyrroline-5-carboxylate; Belongs to the proline oxidase family (600 aa)
PNMA1	Paraneoplastic antigen mal; Paraneoplastic Ma antigens; Belongs to the PNMA family (353 aa)
TOP2A	DNA topoisomerase 2-alpha; Control of topological states of DNA by transient breakage and subsequent rejoining of DNA strands. Topoisomerase II makes double-strand breaks. Essential during mitosis and meiosis for proper segregation of daughter chromosomes. May play a role in regulating the period length of ARNTL/BMAL1 transcriptional oscillation (By similarity); Topoisomerases (1531 aa)
DNAJC7	DnaJ homolog subfamily C member 7; Acts as co-chaperone regulating the molecular chaperones HSP70 and HSP90 in folding of steroid receptors, such as the glucocorticoid receptor and the progesterone receptor. Proposed to act as a recycling chaperone by facilitating the return of chaperone substrates to early stages of chaperoning if further folding is required. In vitro, induces ATP-independent dissociation

	of HSP90 but not of HSP70 from the chaperone- substrate complexes. Recruits NRII3 to the cytoplasm (By similarity); DNAJ heat shock proteins (494 aa)
GP1BA	Platelet glycoprotein Ib alpha chain; GP-Ib, a surface membrane protein of platelets, participates in the formation of platelet plugs by binding to the A1 domain of vWF, which is already bound to the subendothelium; CD molecules (652 aa)
MAP3K1	Mitogen-activated protein kinase kinase kinase 1; Component of a protein kinase signal transduction cascade. Activates the ERK and JNK kinase pathways by phosphorylation of MAP2K1 and MAP2K4. May phosphorylate the MAPK8/JNK1 kinase. Activates CHUK and IKBKB, the central protein kinases of the NF- kappa-B pathway (1512 aa)
NT5C3B	7-methylguanosine phosphate-specific 5'-nucleotidase; Specifically hydrolyzes 7-methylguanosine monophosphate (m(7)GMP) to 7-methylguanosine and inorganic phosphate. The specific activity for m(7)GMP may protect cells against undesired salvage of m(7)GMP and its incorporation into nucleic acids. Also has weak activity for CMP. UMP and purine nucleotides are poor substrates (300 aa)
RANBP1	Ran-specific GTPase-activating protein; Inhibits GTP exchange on Ran. Forms a Ran-GTP-RANBP1 trimeric complex. Increase GTP hydrolysis induced by the Ran GTPase activating protein RANGAP1. May act in an intracellular signaling pathway which may control the progression through the cell cycle by regulating the transport of protein and nucleic acids across the nuclear membrane (278 aa)
CSF3	Granulocyte colony-stimulating factor; Granulocyte/macrophage colony-stimulating factors are cytokines that act in hematopoiesis by controlling the production, differentiation, and function of 2 related white cell populations of the blood, the granulocytes and the monocytes-macrophages. This CSF induces granulocytes; Belongs to the IL-6 superfamily (207 aa)
ERBB2	Receptor tyrosine-protein kinase erbB-2; Protein tyrosine kinase that is part of several cell surface receptor complexes, but that apparently needs a coreceptor for ligand binding. Essential component of a neuregulin-receptor complex, although neuregulins do not interact with it alone. GP30 is a potential ligand for this receptor. Regulates outgrowth and stabilization of peripheral microtubules (MTs). Upon ERBB2 activation, the MEMO1-RHOA-DIAPH1 signaling pathway elicits the phosphorylation and thus the inhibition of GSK3B at cell membrane. This prevents the phosphorylation of APC and [...] (1255 aa)
OR11H12	Olfactory receptor 11H12; Odorant receptor; Olfactory receptors, family 11 (326 aa)
CCDC125	Coiled-coil domain-containing protein 125; May be involved in the regulation of cell migration (511 aa)
SLC6A4	Sodium-dependent serotonin transporter; Serotonin transporter whose primary function in the central nervous system involves the regulation of serotonergic signaling via transport of serotonin molecules from the synaptic cleft back into the pre-synaptic terminal for re-utilization. Plays a key role in mediating regulation of the availability of serotonin to other receptors of serotonergic systems. Terminates the action of serotonin and recycles it in a sodium-dependent manner; Belongs to the sodium:neurotransmitter symporter (SNF) (TC 2.A.22) family. SLC6A4 subfamily (630 aa)
DNAH11	Dynein heavy chain 11, axonemal; Force generating protein of respiratory cilia. Produces force towards the minus ends of microtubules. Dynein has ATPase activity; the force-producing power stroke is thought to occur on release of ADP; Dyneins, axonemal (4516 aa)
DEFB104B	Beta-defensin 104; Has antimicrobial activity. Synergistic effects with lysozyme and DEFB103; Belongs to the beta-defensin family (72 aa)
POTEM	POTE ankyrin domain family member M (508 aa)
CCR7	C-C chemokine receptor type 7; Receptor for the MIP-3-beta chemokine. Probable mediator of EBV effects on B-lymphocytes or of normal lymphocyte functions; Belongs to the G-protein coupled receptor 1 family (378 aa)
LEPREL4	Endoplasmic reticulum protein SC65; Part of a complex composed of PLOD1, P3H3 and P3H4 that catalyzes hydroxylation of lysine residues in collagen alpha chains and is required for normal assembly and cross-linking of collagen fibrils. Required for normal bone density and normal skin stability via its role in hydroxylation of lysine residues in collagen alpha chains and in collagen fibril assembly; Belongs to the leprecan family (437 aa)
VTN	Vitronectin; Vitronectin is a cell adhesion and spreading factor found in serum and tissues. Vitronectin interact with glycosaminoglycans and proteoglycans. Is recognized by certain members of the integrin family and serves as a cell-to-substrate adhesion molecule. Inhibitor of the membrane-damaging effect of the terminal cytolytic complement pathway; Endogenous ligands (478 aa)
MARVELD2	MARVEL domain-containing protein 2; Plays a role in the formation of the epithelial barriers. The separation of the endolymphatic and perilymphatic spaces of the organ of Corti from one another by epithelial barriers is required for normal hearing; Deafness associated genes (558 aa)
PMP22	Peripheral myelin protein 22; Might be involved in growth regulation, and in myelination in the peripheral nervous system; Belongs to the PMP-22/EMP/MP20 family (160 aa)
ZNF705G	Putative zinc finger protein 705G; May be involved in transcriptional regulation; Zinc fingers C2H2-type (300 aa)

AK6	Transcription initiation factor TFIID subunit 9; Broad-specificity nucleoside monophosphate (NMP) kinase that catalyzes the reversible transfer of the terminal phosphate group between nucleoside triphosphates and monophosphates. AMP and dAMP are the preferred substrates, but CMP and dCMP are also good substrates. IMP is phosphorylated to a much lesser extent. All nucleoside triphosphates ATP, GTP, UTP, CTP, dATP, dCTP, dGTP, and TTP are accepted as phosphate donors. CTP is the best phosphate donor, followed by UTP, ATP, GTP and dCTP. May have a role in nuclear energy homeostasis. Has a [...] (264 aa)
FSD1	Fibronectin type III and SPRY domain-containing protein 1; May be involved in microtubule organization and stabilization; Fibronectin type III domain containing (496 aa)
ZMYND10	Zinc finger MYND domain-containing protein 10; Required for motile ciliary function. Probably involved in axonemal assembly of inner and outer dynein arms (IDA and ODA, respectively) for proper axoneme building for cilia motility. May act by indirectly regulating transcription of dynein proteins; Belongs to the ZMYND10 family (440 aa)
CCL5	C-C motif chemokine 5; Chemoattractant for blood monocytes, memory T-helper cells and eosinophils. Causes the release of histamine from basophils and activates eosinophils. May activate several chemokine receptors including CCR1, CCR3, CCR4 and CCR5. One of the major HIV-suppressive factors produced by CD8+ T-cells. Recombinant RANTES protein induces a dose-dependent inhibition of different strains of HIV-1, HIV-2, and simian immunodeficiency virus (SIV). The processed form RANTES(3-68) acts as a natural chemotaxis inhibitor and is a more potent inhibitor of HIV-1- infection. The second [...] (91 aa)
DEFB107B	Beta-defensin 107; Has antibacterial activity; Defensins, beta (70 aa)
RARA	Retinoic acid receptor alpha; Receptor for retinoic acid. Retinoic acid receptors bind as heterodimers to their target response elements in response to their ligands, all-trans or 9-cis retinoic acid, and regulate gene expression in various biological processes. The RXR/RAR heterodimers bind to the retinoic acid response elements (RARE) composed of tandem 5'-AGGTCA-3' sites known as DR1-DR5. In the absence of ligand, the RXR-RAR heterodimers associate with a multiprotein complex containing transcription corepressors that induce histone acetylation, chromatin condensation and transcript [...] (462 aa)
DEFB4B	Beta-defensin 4A; Exhibits antimicrobial activity against Gram-negative bacteria and Gram-positive bacteria. May act as a ligand for C-C chemokine receptor CCR6. Can bind to both human and mouse CCR6 and induce chemotactic activity of CCR6-expressing cells; Belongs to the beta-defensin family. LAP/TAP subfamily (64 aa)
NKIRAS2	NF-kappa-B inhibitor-interacting Ras-like protein 2; Atypical Ras-like protein that acts as a potent regulator of NF-kappa-B activity by preventing the degradation of NF-kappa-B inhibitor beta (NFKBIB) by most signals, explaining why NFKBIB is more resistant to degradation. May act by blocking phosphorylation of NFKBIB and nuclear localization of p65/RELA NF- kappa-B subunit. It is unclear whether it acts as a GTPase. Both GTP- and GDP-bound forms block phosphorylation of NFKBIB (By similarity); Belongs to the small GTPase superfamily. Ras family. KappaB-Ras subfamily (191 aa)
DEFB106B	Defensin, beta 106B; Has antibacterial activity. Acts as a ligand for C-C chemokine receptor CCR2; Belongs to the beta-defensin family (65 aa)
ZFPM2	Zinc finger protein ZFPM2; Transcription regulator that plays a central role in heart morphogenesis and development of coronary vessels from epicardium, by regulating genes that are essential during cardiogenesis. Essential cofactor that acts via the formation of a heterodimer with transcription factors of the GATA family GATA4, GATA5 and GATA6. Such heterodimer can both activate or repress transcriptional activity, depending on the cell and promoter context. Also required in gonadal differentiation, possibly by regulating expression of SRY. Probably acts as a corepressor of NR2F2 (By similarity) [...] (1151 aa)
OCLN	Occludin; May play a role in the formation and regulation of the tight junction (TJ) paracellular permeability barrier. It is able to induce adhesion when expressed in cells lacking tight junctions; Protein phosphatase 1 regulatory subunits (522 aa)
RBFOX1	RNA binding protein fox-1 homolog 1; RNA-binding protein that regulates alternative splicing events by binding to 5'-UGCAUGU-3' elements. Regulates alternative splicing of tissue-specific exons and of differentially spliced exons during erythropoiesis (418 aa)
POLR2A	DNA-directed RNA polymerase II subunit RPB1; DNA-dependent RNA polymerase catalyzes the transcription of DNA into RNA using the four ribonucleoside triphosphates as substrates. Largest and catalytic component of RNA polymerase II which synthesizes mRNA precursors and many functional non-coding RNAs. Forms the polymerase active center together with the second largest subunit. Pol II is the central component of the basal RNA polymerase II transcription machinery. It is composed of mobile elements that move relative to each other. RPB1 is part of the core element with the central large c1 [...] (1980 aa)
DNAH5	Dynein heavy chain 5, axonemal; Force generating protein of respiratory cilia. Produces force towards the minus ends of microtubules. Dynein has ATPase activity; the force-producing power stroke is thought

	to occur on release of ADP. Required for structural and functional integrity of the cilia of ependymal cells lining the brain ventricles; Dyneins, axonemal (4624 aa)
SUN3	Sad1 and UNC84 domain containing 3; SUN domain-containing protein 3; As a probable component of the LINC (LInker of Nucleoskeleton and Cytoskeleton) complex, involved in the connection between the nuclear lamina and the cytoskeleton. The nucleocytoplasmic interactions established by the LINC complex play an important role in the transmission of mechanical forces across the nuclear envelope and in nuclear movement and positioning. May be involved in nuclear remodeling during sperm head formation in spermatogenesis. A probable SUN3:SYNE1 LINC complex may tether spermatid nuclei to poster [...] (357 aa)
TRMT2A	tRNA (uracil-5-)-methyltransferase homolog A; May be involved in nucleic acid metabolism and/or modifications; tRNA methyltransferases (625 aa)
CCL2	C-C motif chemokine 2; Chemotactic factor that attracts monocytes and basophils but not neutrophils or eosinophils. Augments monocyte anti-tumor activity. Has been implicated in the pathogenesis of diseases characterized by monocytic infiltrates, like psoriasis, rheumatoid arthritis or atherosclerosis. May be involved in the recruitment of monocytes into the arterial wall during the disease process of atherosclerosis; Belongs to the intercrine beta (chemokine CC) family (99 aa)
ACLY	ATP-citrate synthase; ATP-citrate synthase is the primary enzyme responsible for the synthesis of cytosolic acetyl-CoA in many tissues. Has a central role in de novo lipid synthesis. In nervous tissue it may be involved in the biosynthesis of acetylcholine; In the N-terminal section; belongs to the succinate/malate CoA ligase beta subunit family (1101 aa)
PIH1D3	Protein PIH1D3; Plays a role in cytoplasmic pre-assembly of axonemal dynein; Belongs to the PIH1 family (214 aa)
FKBP10	Peptidyl-prolyl cis-trans isomerase FKBP10; PPIases accelerate the folding of proteins during protein synthesis; EF-hand domain containing (582 aa)
CDK11B	Cyclin-dependent kinase 11B; Plays multiple roles in cell cycle progression, cytokinesis and apoptosis. Involved in pre-mRNA splicing in a kinase activity-dependent manner. Isoform 7 may act as a negative regulator of normal cell cycle progression; Cyclin dependent kinases (795 aa)
MMP23B	Matrix metalloproteinase-23; Protease. May regulate the surface expression of some potassium channels by retaining them in the endoplasmic reticulum (By similarity); Immunoglobulin like domain containing (390 aa)
MYO1D	Unconventional myosin-Id; Myosins are actin-based motor molecules with ATPase activity. Unconventional myosins serve in intracellular movements. Their highly divergent tails are presumed to bind to membranous compartments, which would be moved relative to actin filaments (By similarity); Myosins, class I (1006 aa)
PDXDC1	Pyridoxal-dependent decarboxylase domain-containing protein 1; Pyridoxal dependent decarboxylase domain containing 1; Belongs to the group II decarboxylase family (788 aa)
NF1	Neurofibromin; Stimulates the GTPase activity of Ras. NF1 shows greater affinity for Ras GAP, but lower specific activity. May be a regulator of Ras activity; Armadillo-like helical domain containing (2839 aa)
JUP	Junction plakoglobin; Common junctional plaque protein. The membrane-associated plaques are architectural elements in an important strategic position to influence the arrangement and function of both the cytoskeleton and the cells within the tissue. The presence of plakoglobin in both the desmosomes and in the intermediate junctions suggests that it plays a central role in the structure and function of submembranous plaques. Acts as a substrate for VE-PTP and is required by it to stimulate VE-cadherin function in endothelial cells. Can replace beta-catenin in E-cadherin/catenin adhes [...] (745 aa)
SOX9	Transcription factor SOX-9; Transcriptional regulator. Binds to the COL2A1 promoter and activates COL2A1 expression, as part of a complex with ZNF219 (By similarity). Plays a role in chondrocyte differentiation (By similarity). Important for normal skeletal development; SRY-boxes (509 aa)
DGCR6	DiGeorge syndrome critical region gene 6; May play a role in neural crest cell migration into the third and fourth pharyngeal pouches; Belongs to the gonadal family (220 aa)
NCOR1	Nuclear receptor co-repressor 1; Nuclear receptor corepressor 1; Mediates transcriptional repression by certain nuclear receptors. Part of a complex which promotes histone deacetylation and the formation of repressive chromatin structures which may impede the access of basal transcription factors. Participates in the transcriptional repressor activity produced by BCL6 (2440 aa)
DEFB103B	Beta-defensin 103; Exhibits antimicrobial activity against Gram-positive bacteria S.aureus and S.pyogenes, Gram-negative bacteria P.aeruginosa and E.coli and the yeast C.albicans. Kills multiresistant S.aureus and vancomycin-resistant E.faecium. No significant hemolytic activity was observed; Defensins, beta (67 aa)
DEFB105B	Beta-defensin 105; Has antibacterial activity; Defensins, beta (78 aa)

DNAAF2	Dynein assembly factor 2, axonemal; Protein kintoun; Required for cytoplasmic pre-assembly of axonemal dyneins, thereby playing a central role in motility in cilia and flagella. Involved in pre-assembly of dynein arm complexes in the cytoplasm before intraflagellar transport loads them for the ciliary compartment (837 aa)
SSU72	RNA polymerase II subunit A C-terminal domain phosphatase SSU72; Protein phosphatase that catalyzes the dephosphorylation of the C-terminal domain of RNA polymerase II. Plays a role in RNA processing and termination. Plays a role in pre-mRNA polyadenylation via its interaction with SYMPK; Belongs to the SSU72 phosphatase family (194 aa)
HYDIN	Hydin, axonemal central pair apparatus protein; Hydrocephalus-inducing protein homolog; Required for ciliary motility; Protein phosphatase 1 regulatory subunits (5121 aa)
CUX1	Homeobox protein cut-like 1; Probably has a broad role in mammalian development as a repressor of developmentally regulated gene expression. May act by preventing binding of positively-activating CCAAT factors to promoters. Component of nf-munr repressor; binds to the matrix attachment regions (MARs) (5' and 3') of the immunoglobulin heavy chain enhancer. Represses T-cell receptor (TCR) beta enhancer function by binding to MARbeta, an ATC-rich DNA sequence located upstream of the TCR beta enhancer. Binds to the TH enhancer; may require the basic helix-loop-helix protein TCF4 as a coactiv [...] (1516 aa)
ALOX15	Arachidonate 15-lipoxygenase; Non-heme iron-containing dioxygenase that catalyzes the stereo-specific peroxidation of free and esterified polyunsaturated fatty acids generating a spectrum of bioactive lipid mediators. Converts arachidonic acid into 12- hydroperoxyeicosatetraenoic acid/12-HPETE and 15-hydroperoxyeicosatetraenoic acid/15-HPETE. Also converts linoleic acid to 13-hydroperoxyoctadecadienoic acid. May also act on (12S)- hydroperoxyeicosatetraenoic acid/(12S)-HPETE to produce hepoxilin A3. Probably plays an important role in the immune and inflammatory responses. Through the [...] (662 aa)
TNFSF13	Tumor necrosis factor ligand superfamily member 13; Cytokine that binds to TNFRSF13B/TACI and to TNFRSF17/BCMA. Plays a role in the regulation of tumor cell growth. May be involved in monocyte/macrophage-mediated immunological processes; CD molecules (250 aa)
ZIC3	Zinc finger protein ZIC 3; Acts as transcriptional activator. Required in the earliest stages in both axial midline development and left-right (LR) asymmetry specification. Binds to the minimal GLI-consensus sequence 5'-GGGTGGTC-3'; Belongs to the GLI C2H2-type zinc-finger protein family (467 aa)
MIB2	E3 ubiquitin-protein ligase MIB2; E3 ubiquitin-protein ligase that mediates ubiquitination of Delta receptors, which act as ligands of Notch proteins. Positively regulates the Delta-mediated Notch signaling by ubiquitinating the intracellular domain of Delta, leading to endocytosis of Delta receptors; Ankyrin repeat domain containing (1070 aa)
ZDHHC8	Probable palmitoyltransferase ZDHHC8; Palmitoyltransferase involved in glutamatergic transmission. Mediates palmitoylation of ABCA1; Belongs to the DHHC palmitoyltransferase family. ERF2/ZDHHC9 subfamily (778 aa)
CDH3	Cadherin-3; Cadherins are calcium-dependent cell adhesion proteins. They preferentially interact with themselves in a homophilic manner in connecting cells; cadherins may thus contribute to the sorting of heterogeneous cell types (829 aa)
USP17L4	Inactive ubiquitin carboxyl-terminal hydrolase 17-like protein 4; Ubiquitin specific peptidase 17-like family member 4 (530 aa)
DNAH1	Dynein heavy chain 1, axonemal; Force generating protein of cilia required for sperm flagellum motility. Produces force towards the minus ends of microtubules. Dynein has ATPase activity; the force-producing power stroke is thought to occur on release of ADP. Required in spermatozoa for the formation of the inner dynein arms and biogenesis of the axoneme; Belongs to the dynein heavy chain family (4265 aa)
DNAAF3	Dynein assembly factor 3, axonemal; Required for the assembly of axonemal inner and outer dynein arms. Involved in preassembly of dyneins into complexes before their transport into cilia; Belongs to the DNAAF3 family (608 aa)
SRXN1	Sulfiredoxin-1; Contributes to oxidative stress resistance by reducing cysteine-sulfinic acid formed under exposure to oxidants in the peroxiredoxins PRDX1, PRDX2, PRDX3 and PRDX4. Does not act on PRDX5 or PRDX6. May catalyze the reduction in a multi-step process by acting both as a specific phosphotransferase and a thioltransferase; Belongs to the sulfiredoxin family (137 aa)
CECR1	Adenosine deaminase cecr1; Adenosine deaminase 2; Adenosine deaminase that may contribute to the degradation of extracellular adenosine, a signaling molecule that controls a variety of cellular responses. Requires elevated adenosine levels for optimal enzyme activity. Binds to cell surfaces via proteoglycans and may play a role in the regulation of cell proliferation and differentiation, independently of its enzyme activity (511 aa)
SHBG	Sex hormone-binding globulin; Functions as an androgen transport protein, but may also be involved in receptor mediated processes. Each dimer binds one molecule of steroid. Specific for 5-alpha-

	dihydrotestosterone, testosterone, and 17-beta-estradiol. Regulates the plasma metabolic clearance rate of steroid hormones by controlling their plasma concentration (402 aa)
TTC25	Tetratricopeptide repeat protein 25; Required for the docking of the outer dynein arm to cilia, hence plays an essential role in cilia motility; Tetratricopeptide repeat domain containing (672 aa)
CBX2	Chromobox protein homolog 2; Component of a Polycomb group (PcG) multiprotein PRC1- like complex, a complex class required to maintain the transcriptionally repressive state of many genes, including Hox genes, throughout development. PcG PRC1 complex acts via chromatin remodeling and modification of histones; it mediates monoubiquitination of histone H2A 'Lys-119', rendering chromatin heritably changed in its expressibility. Binds to histone H3 trimethylated at 'Lys-9' (H3K9me3) or at 'Lys-27' (H3K27me3) (By similarity). Plays a role in the lineage differentiation of the germ layers in [...] (532 aa)
RAD17	Cell cycle checkpoint protein RAD17; Essential for sustained cell growth, maintenance of chromosomal stability, and ATR-dependent checkpoint activation upon DNA damage. Has a weak ATPase activity required for binding to chromatin. Participates in the recruitment of the RAD1-RAD9- HUS1 complex and RHNO1 onto chromatin, and in CHEK1 activation. May also serve as a sensor of DNA replication progression, and may be involved in homologous recombination; Belongs to the rad17/RAD24 family (681 aa)
DGCR8	DGCR8, microprocessor complex subunit; Microprocessor complex subunit DGCR8; Component of the microprocessor complex that acts as a RNA- and heme-binding protein that is involved in the initial step of microRNA (miRNA) biogenesis. Component of the microprocessor complex that is required to process primary miRNA transcripts (pri-miRNAs) to release precursor miRNA (pre-miRNA) in the nucleus. Within the microprocessor complex, DGCR8 function as a molecular anchor necessary for the recognition of pri-miRNA at dsRNA-ssRNA junction and directs DROSHA to cleave 11 bp away form the junction to [...] (773 aa)
DHX58	Probable ATP-dependent RNA helicase DHX58; Acts as a regulator of DDX58/RIG-I and IFIH1/MDA5 mediated antiviral signaling. Cannot initiate antiviral signaling as it lacks the CARD domain required for activating MAVS/IPS1- dependent signaling events. Can have both negative and positive regulatory functions related to DDX58/RIG-I and IFIH1/MDA5 signaling and this role in regulating signaling may be complex and could probably depend on characteristics of the infecting virus or target cells, or both. Its inhibitory action on DDX58/RIG-I signaling may involve the following mechanisms: compe [...] (678 aa)
ZNF385C	Zinc finger protein 385C (424 aa)
PNMA6A	Paraneoplastic antigen-like protein 6A; Paraneoplastic Ma antigens; Belongs to the PNMA family (399 aa)
TP53	Cellular tumor antigen p53; Acts as a tumor suppressor in many tumor types; induces growth arrest or apoptosis depending on the physiological circumstances and cell type. Involved in cell cycle regulation as a trans-activator that acts to negatively regulate cell division by controlling a set of genes required for this process. One of the activated genes is an inhibitor of cyclin-dependent kinases. Apoptosis induction seems to be mediated either by stimulation of BAX and FAS antigen expression, or by repression of Bcl-2 expression. In cooperation with mitochondrial PPIF is involved in [...] (393 aa)
BFSP2	Beaded filament structural protein 2; Phakinin; Involved in stabilization of lens fiber cell cytoskeleton; Belongs to the intermediate filament family (415 aa)
TNFSF12	Tumor necrosis factor ligand superfamily member 12; Binds to FN14 and possibly also to TNRFSF12/APO3. Weak inducer of apoptosis in some cell types. Mediates NF-kappa-B activation. Promotes angiogenesis and the proliferation of endothelial cells. Also involved in induction of inflammatory cytokines. Promotes IL8 secretion; Tumor necrosis factor superfamily (249 aa)
KAT2A	Histone acetyltransferase KAT2A; Functions as a histone acetyltransferase (HAT) to promote transcriptional activation. Acetylation of histones gives a specific tag for epigenetic transcription activation. Has significant histone acetyltransferase activity with core histones, but not with nucleosome core particles. Also acetylates non- histone proteins, such as CEBPB. Component of the ATAC complex, a complex with histone acetyltransferase activity on histones H3 and H4. In case of HIV-1 infection, it is recruited by the viral protein Tat. Regulates Tat's transactivating activity and may [...] (837 aa)
KRT19	Keratin, type I cytoskeletal 19; Involved in the organization of myofibers. Together with KRT8, helps to link the contractile apparatus to dystrophin at the costameres of striated muscle; Keratins, type I (400 aa)
SNX18	Sorting nexin-18; Involved in endocytosis and intracellular vesicle trafficking, both during interphase and at the end of mitosis. Required for efficient progress through mitosis and cytokinesis. Required for normal formation of the cleavage furrow at the end of mitosis. Plays a role in endocytosis via clathrin-coated pits, but also clathrin-independent, actin-dependent fluid-phase endocytosis. Plays a role in macropinocytosis. Binds to membranes enriched in phosphatidylinositol 4,5-bisphosphate and promotes membrane tubulation. Stimulates the GTPase activity of DNM2. Promotes DNM2 loc [...] (628 aa)

DNAI1	Dynein intermediate chain 1, axonemal; Part of the dynein complex of respiratory cilia; Belongs to the dynein intermediate chain family (703 aa)
HNF1B	Hepatocyte nuclear factor 1-beta; Transcription factor, probably binds to the inverted palindrome 5'-GTTAATNATTAAC-3'; Belongs to the HNF1 homeobox family (557 aa)
NR0B1	Nuclear receptor subfamily 0 group B member 1; Orphan nuclear receptor. Component of a cascade required for the development of the hypothalamic-pituitary-adrenal-gonadal axis. Acts as a coregulatory protein that inhibits the transcriptional activity of other nuclear receptors through heterodimeric interactions. May also have a role in the development of the embryo and in the maintenance of embryonic stem cell pluripotency (By similarity); Belongs to the nuclear hormone receptor family. NR0 subfamily (470 aa)
SLC2A4	Solute carrier family 2, facilitated glucose transporter member 4; Insulin-regulated facilitative glucose transporter; Solute carriers (509 aa)
FAM115C	TRPM8 channel-associated factor 2; Isoform 2: Negatively regulates the plasma membrane cation channel TRPM8 activity. Involved in the recruitment of TRPM8 to the cell surface. Promotes prostate cancer cell migration stimulation in a TRPM8-dependent manner (845 aa)
AKR1C2	3alpha-hydroxysteroid 3-dehydrogenase; Aldo-keto reductase family 1 member C2; Works in concert with the 5-alpha/5-beta-steroid reductases to convert steroid hormones into the 3-alpha/5-alpha and 3-alpha/5-beta-tetrahydrosteroids. Catalyzes the inactivation of the most potent androgen 5-alpha-dihydrotestosterone (5-alpha-DHT) to 5-alpha-androstane-3-alpha,17-beta-diol (3-alpha-diol). Has a high bile-binding ability (323 aa)
TAF9	Tata-box binding protein associated factor 9; Broad-specificity nucleoside monophosphate (NMP) kinase that catalyzes the reversible transfer of the terminal phosphate group between nucleoside triphosphates and monophosphates. AMP and dAMP are the preferred substrates, but CMP and dCMP are also good substrates. IMP is phosphorylated to a much lesser extent. All nucleoside triphosphates ATP, GTP, UTP, CTP, dATP, dCTP, dGTP, and TTP are accepted as phosphate donors. CTP is the best phosphate donor, followed by UTP, ATP, GTP and dCTP. May have a role in nuclear energy homeostasis. Has also [...] (172 aa)
C1orf233	Fibronectin type III domain-containing protein 10; Chromosome 1 open reading frame 233 (226 aa)
ARRB2	Beta-arrestin-2; Functions in regulating agonist-mediated G-protein coupled receptor (GPCR) signaling by mediating both receptor desensitization and resensitization processes. During homologous desensitization, beta-arrestins bind to the GPRK-phosphorylated receptor and sterically preclude its coupling to the cognate G- protein; the binding appears to require additional receptor determinants exposed only in the active receptor conformation. The beta-arrestins target many receptors for internalization by acting as endocytic adapters (CLASPs, clathrin-associated sorting proteins) and rec [...] (430 aa)
AURKB	Aurora kinase B; Serine/threonine-protein kinase component of the chromosomal passenger complex (CPC), a complex that acts as a key regulator of mitosis. The CPC complex has essential functions at the centromere in ensuring correct chromosome alignment and segregation and is required for chromatin-induced microtubule stabilization and spindle assembly. Involved in the bipolar attachment of spindle microtubules to kinetochores and is a key regulator for the onset of cytokinesis during mitosis. Required for central/midzone spindle assembly and cleavage furrow formation. Key component of [...] (345 aa)
DAB1	DAB1, reelin adaptor protein; Disabled homolog 1; Adapter molecule functioning in neural development. May regulate SIAH1 activity (555 aa)
CCDC40	Coiled-coil domain-containing protein 40; Required for assembly of dynein regulatory complex (DRC) and inner dynein arm (IDA) complexes, which are responsible for ciliary beat regulation, thereby playing a central role in motility in cilia and flagella. Probably acts together with CCDC39 to form a molecular ruler that determines the 96 nanometer (nm) repeat length and arrangements of components in cilia and flagella (By similarity). Not required for outer dynein arm complexes assembly. Required for axonemal recruitment of CCDC39; Belongs to the CCDC40 family (1142 aa)
TANGO2	Transport and golgi organization 2 homolog (317 aa)
SREBF1	Sterol regulatory element-binding protein 1; Transcriptional activator required for lipid homeostasis. Regulates transcription of the LDL receptor gene as well as the fatty acid and to a lesser degree the cholesterol synthesis pathway (By similarity). Binds to the sterol regulatory element 1 (SRE-1) (5'-ATCACCCAC-3'). Has dual sequence specificity binding to both an E-box motif (5'-ATCACGTGA-3') and to SRE-1 (5'-ATCACCCAC-3'); Basic helix-loop-helix proteins (1177 aa)
CNP	2',3'-cyclic-nucleotide 3'-phosphodiesterase; May participate in RNA metabolism in the myelinating cell, CNP is the third most abundant protein in central nervous system myelin (421 aa)