

**Supplementary information, Table S16** Functional enrichment categories of PSGs reported by Rubin and colleagues in layer lines(LR). Categories associated with vision-related function are marked in green.

Term	Term ID	P-value	Gene number	Descriptions
ke	KEGG:04020	5.00E-02	7	Calcium signaling pathway
hp	HP:0100699	4.36E-02	2	Scarring
hp	HP:0100689	2.51E-02	2	Decreased corneal thickness
hp	HP:0100659	1.15E-02	3	Abnormality of the cerebral vasculature
hp	HP:0100491	1.76E-02	5	Abnormality of lower limb joint
hp	HP:0040195	3.67E-02	7	Decreased head circumference
hp	HP:0030453	1.00E-02	4	Abnormal visual electrophysiology
hp	HP:0030319	8.98E-03	3	Weakness of facial musculature
hp	HP:0030311	2.69E-02	2	Lower extremity joint dislocation
hp	HP:0030178	8.04E-03	4	Abnormality of central nervous system electrophysiology
hp	HP:0012823	3.85E-02	11	Clinical modifier
hp	HP:0012758	3.58E-02	10	Neurodevelopmental delay
hp	HP:0012547	4.54E-02	7	Abnormal involuntary eye movements
hp	HP:0012503	3.29E-02	2	Abnormality of the pituitary gland
hp	HP:0012387	4.48E-03	2	Bronchitis
hp	HP:0012265	3.63E-03	2	Ciliary dyskinesia
hp	HP:0012262	3.91E-03	2	Abnormal ciliary motility
hp	HP:0012261	3.91E-03	2	Abnormal respiratory motile cilium physiology
hp	HP:0012223	4.29E-05	2	Splenic rupture
hp	HP:0011900	2.91E-04	2	Hypofibrinogenemia
hp	HP:0011898	2.91E-04	2	Abnormality of circulating fibrinogen
hp	HP:0011843	4.13E-02	4	Abnormality of skeletal physiology
hp	HP:0011842	8.18E-03	22	Abnormality of skeletal morphology
hp	HP:0011830	3.88E-02	2	Abnormality of oral mucosa
hp	HP:0011821	1.87E-02	9	Abnormality of facial skeleton
hp	HP:0011799	2.42E-02	3	Abnormality of facial soft tissue
hp	HP:0011747	3.15E-02	2	Abnormality of the anterior pituitary
hp	HP:0011733	1.96E-02	2	Abnormality of adrenal physiology
hp	HP:0011675	3.43E-02	4	Arrhythmia
hp	HP:0011492	4.31E-02	2	Abnormality of corneal stroma
hp	HP:0011486	2.60E-02	2	Abnormality of corneal thickness
hp	HP:0011463	9.18E-03	2	Childhood onset
hp	HP:0011389	1.66E-02	8	Functional abnormality of the inner ear
hp	HP:0011198	6.35E-03	2	EEG with generalized epileptiform discharges
hp	HP:0011182	6.69E-03	2	Epileptiform EEG discharges

hp	HP:0011039	1.96E-02	2	Abnormality of the helix
hp	HP:0011029	4.23E-02	2	Internal hemorrhage
hp	HP:0011028	4.23E-02	2	Abnormality of blood circulation
hp	HP:0011025	6.49E-03	10	Abnormality of cardiovascular system physiology
hp	HP:0010991	1.44E-02	2	Abnormality of the abdominal musculature
hp	HP:0010990	1.56E-03	2	Abnormality of the common coagulation pathway
hp	HP:0010827	8.74E-03	3	Abnormality of the seventh cranial nerve
hp	HP:0010696	3.11E-03	2	Polar cataract
hp	HP:0010628	8.74E-03	3	Facial palsy
hp	HP:0010549	4.66E-02	3	Weakness due to upper motor neuron dysfunction
hp	HP:0010490	3.24E-02	2	Abnormality of the palmar creases
hp	HP:0010460	4.34E-02	5	Abnormality of the female genitalia
hp	HP:0010318	1.11E-02	2	Aplasia/Hypoplasia of the abdominal wall musculature
hp	HP:0009122	3.44E-02	6	Aplasia/hypoplasia affecting bones of the axial skeleton
hp	HP:0009121	1.39E-02	18	Abnormal axial skeleton morphology
hp	HP:0009118	2.32E-02	6	Aplasia/Hypoplasia of the mandible
hp	HP:0009116	2.42E-02	6	Aplasia/Hypoplasia involving bones of the skull
hp	HP:0008736	2.61E-02	4	Hypoplasia of penis
hp	HP:0008678	4.82E-02	2	Renal hypoplasia/aplasia
hp	HP:0008373	4.59E-02	3	Puberty and gonadal disorders
hp	HP:0008064	3.43E-02	2	Ichthyosis
hp	HP:0008048	2.17E-03	2	Abnormality of the line of Schwalbe
hp	HP:0008047	3.97E-02	2	Abnormality of the vasculature of the eye
hp	HP:0008046	3.61E-02	2	Abnormality of the retinal vasculature
hp	HP:0008034	1.48E-02	2	Abnormal iris pigmentation
hp	HP:0007759	4.27E-02	2	Opacification of the corneal stroma
hp	HP:0007703	3.69E-02	4	Abnormality of retinal pigmentation
hp	HP:0007663	8.74E-03	3	Reduced visual acuity
hp	HP:0007477	4.49E-02	2	Abnormal dermatoglyphics
hp	HP:0006824	1.93E-02	3	Cranial nerve paralysis
hp	HP:0006703	2.23E-02	2	Aplasia/Hypoplasia of the lungs
hp	HP:0005978	2.78E-02	2	Type II diabetes mellitus
hp	HP:0005268	1.03E-03	2	Spontaneous abortion
hp	HP:0005262	1.26E-02	3	Abnormality of the synovia
hp	HP:0004936	7.72E-03	2	Venous thrombosis
hp	HP:0004418	3.37E-03	2	Thrombophlebitis
hp	HP:0004374	4.82E-02	2	Hemiplegia/hemiparesis

hp	HP:0004348	3.14E-02	5	Abnormality of bone mineral density
hp	HP:0004328	1.13E-02	11	Abnormality of the anterior segment of the eye
hp	HP:0004324	2.20E-02	4	Increased body weight
hp	HP:0003828	4.31E-02	2	Variable expressivity
hp	HP:0003745	1.31E-02	2	Sporadic
hp	HP:0003674	3.59E-02	8	Onset
hp	HP:0003577	4.61E-02	2	Congenital onset
hp	HP:0003457	3.65E-02	2	EMG abnormality
hp	HP:0003256	9.18E-03	2	Abnormality of the coagulation cascade
hp	HP:0003241	3.17E-02	4	External genital hypoplasia
hp	HP:0003117	2.42E-02	3	Abnormality of circulating hormone level
hp	HP:0002837	1.56E-03	2	Recurrent bronchitis
hp	HP:0002827	2.05E-02	2	Hip dislocation
hp	HP:0002817	4.24E-02	9	Abnormality of the upper limb
hp	HP:0002815	2.75E-02	3	Abnormality of the knees
hp	HP:0002808	3.75E-02	3	Kyphosis
hp	HP:0002804	1.57E-02	2	Arthrogryposis multiplex congenita
hp	HP:0002803	1.92E-02	2	Congenital contracture
hp	HP:0002795	1.65E-02	8	Functional respiratory abnormality
hp	HP:0002788	3.67E-03	3	Recurrent upper respiratory tract infections
hp	HP:0002750	4.70E-02	2	Delayed skeletal maturation
hp	HP:0002652	4.01E-02	2	Skeletal dysplasia
hp	HP:0002539	1.37E-03	2	Cortical dysplasia
hp	HP:0002538	3.79E-02	2	Abnormality of the cerebral cortex
hp	HP:0002521	6.02E-03	2	Hypsarrhythmia
hp	HP:0002510	8.81E-03	2	Spastic tetraplegia
hp	HP:0002360	3.74E-02	2	Sleep disturbance
hp	HP:0002353	6.27E-03	4	EEG abnormality
hp	HP:0002269	1.40E-02	4	Abnormality of neuronal migration
hp	HP:0002239	2.37E-02	2	Gastrointestinal hemorrhage
hp	HP:0002170	9.94E-03	2	Intracranial hemorrhage
hp	HP:0002110	8.08E-03	2	Bronchiectasis
hp	HP:0002109	1.83E-02	2	Abnormality of the bronchi
hp	HP:0002093	1.86E-02	6	Respiratory insufficiency
hp	HP:0002089	1.31E-02	2	Pulmonary hypoplasia
hp	HP:0002071	3.11E-02	2	Abnormality of extrapyramidal motor function
hp	HP:0001999	4.07E-02	6	Abnormal facial shape
hp	HP:0001977	1.27E-02	2	Abnormal thrombosis
hp	HP:0001933	4.01E-02	2	Subcutaneous hemorrhage
hp	HP:0001928	4.87E-03	4	Abnormality of coagulation
hp	HP:0001892	1.09E-02	5	Abnormal bleeding
hp	HP:0001850	2.19E-02	2	Abnormality of the tarsal bones
hp	HP:0001787	4.19E-03	2	Abnormal delivery

hp	HP:0001761	4.78E-02	2	Pes cavus
hp	HP:0001739	5.56E-03	3	Abnormality of the nasopharynx
hp	HP:0001710	1.87E-02	2	Conotruncal defect
hp	HP:0001695	2.78E-02	2	Cardiac arrest
hp	HP:0001649	4.31E-03	3	Tachycardia
hp	HP:0001645	2.65E-02	2	Sudden cardiac death
hp	HP:0001639	3.95E-02	3	Hypertrophic cardiomyopathy
hp	HP:0001636	1.83E-02	2	Tetralogy of Fallot
hp	HP:0001626	3.05E-02	16	Abnormality of the cardiovascular system
hp	HP:0001622	5.37E-03	3	Premature birth
hp	HP:0001608	1.00E-02	4	Abnormality of the voice
hp	HP:0001561	2.88E-02	2	Polyhydramnios
hp	HP:0001513	1.92E-02	4	Obesity
hp	HP:0001399	4.44E-02	2	Hepatic failure
hp	HP:0001386	3.37E-03	2	Joint swelling
hp	HP:0001384	1.20E-02	3	Abnormality of the hip joint
hp	HP:0001373	4.98E-02	2	Joint dislocation
hp	HP:0001367	3.29E-02	10	Abnormal joint morphology
hp	HP:0001347	4.16E-02	5	Hyperreflexia
hp	HP:0001324	4.91E-02	6	Muscle weakness
hp	HP:0001319	1.96E-02	2	Neonatal hypotonia
hp	HP:0001311	2.15E-02	4	Abnormal nervous system electrophysiology
hp	HP:0001291	2.38E-02	3	Abnormality of the cranial nerves
hp	HP:0001284	3.83E-02	2	Areflexia
hp	HP:0001279	2.62E-03	2	Syncope
hp	HP:0001270	1.29E-02	4	Motor delay
hp	HP:0001268	4.57E-02	2	Mental deterioration
hp	HP:0001256	1.74E-02	2	Intellectual disability, mild
hp	HP:0001250	3.80E-02	11	Seizures
hp	HP:0001249	2.65E-02	10	Intellectual disability
hp	HP:0001197	1.13E-02	6	Abnormality of prenatal development or birth
hp	HP:0001018	3.24E-02	2	Abnormal palmar dermatoglyphics
hp	HP:0000987	4.18E-02	2	Atypical scarring of skin
hp	HP:0000954	2.55E-02	2	Single transverse palmar crease
hp	HP:0000929	1.75E-02	15	Abnormality of the skull
hp	HP:0000927	2.75E-02	3	Abnormality of skeletal maturation
hp	HP:0000924	1.40E-02	22	Abnormality of the skeletal system
hp	HP:0000858	2.00E-02	3	Menstrual irregularities
hp	HP:0000842	2.60E-02	2	Hyperinsulinemia
hp	HP:0000834	3.34E-02	2	Abnormality of the adrenal glands
hp	HP:0000822	1.20E-02	4	Hypertension
hp	HP:0000819	3.71E-02	3	Diabetes mellitus
hp	HP:0000818	8.34E-03	11	Abnormality of the endocrine system

hp	HP:0000752	1.17E-02	3	Hyperactivity
hp	HP:0000750	2.52E-02	3	Delayed speech and language development
hp	HP:0000729	2.92E-02	2	Autistic behavior
hp	HP:0000708	4.26E-02	8	Behavioral abnormality
hp	HP:0000662	1.81E-02	3	Nyctalopia
hp	HP:0000639	4.49E-02	7	Nystagmus
hp	HP:0000627	1.95E-03	2	Posterior embryotoxon
hp	HP:0000613	4.53E-02	2	Photophobia
hp	HP:0000600	8.28E-03	3	Abnormality of the pharynx
hp	HP:0000598	3.21E-02	14	Abnormality of the ear
hp	HP:0000597	2.65E-02	3	Ophthalmoparesis
hp	HP:0000580	1.04E-02	4	Pigmentary retinopathy
hp	HP:0000556	1.31E-02	4	Retinal dystrophy
hp	HP:0000525	4.63E-02	3	Abnormality of the iris
hp	HP:0000518	3.56E-02	6	Cataract
hp	HP:0000517	2.26E-02	7	Abnormality of the lens
hp	HP:0000512	5.97E-03	4	Abnormal electroretinogram
hp	HP:0000510	3.14E-03	4	Rod-cone dystrophy
hp	HP:0000508	3.04E-02	5	Ptosis
hp	HP:0000505	2.49E-02	7	Visual impairment
hp	HP:0000504	4.47E-02	7	Abnormality of vision
hp	HP:0000501	7.52E-03	5	Glaucoma
hp	HP:0000481	4.13E-02	5	Abnormality of the cornea
hp	HP:0000421	1.20E-03	3	Epistaxis
hp	HP:0000407	2.60E-02	7	Sensorineural hearing impairment
hp	HP:0000403	6.35E-03	2	Recurrent otitis media
hp	HP:0000388	3.38E-02	2	Otitis media
hp	HP:0000370	3.20E-02	4	Abnormality of the middle ear
hp	HP:0000368	4.82E-02	3	Low-set, posteriorly rotated ears
hp	HP:0000366	2.73E-02	11	Abnormality of the nose
hp	HP:0000359	1.71E-02	8	Abnormality of the inner ear
hp	HP:0000358	3.46E-02	4	Posteriorly rotated ears
hp	HP:0000347	2.30E-02	6	Micrognathia
hp	HP:0000301	1.04E-02	3	Abnormality of facial musculature
hp	HP:0000280	4.01E-02	2	Coarse facial features
hp	HP:0000277	3.96E-02	6	Abnormality of the mandible
hp	HP:0000271	1.87E-02	19	Abnormality of the face
hp	HP:0000268	3.20E-02	2	Dolichocephaly
hp	HP:0000252	3.67E-02	7	Microcephaly
hp	HP:0000246	2.19E-02	2	Sinusitis
hp	HP:0000245	2.69E-02	2	Abnormality of the paranasal sinuses
hp	HP:0000240	3.75E-02	9	Abnormality of skull size
hp	HP:0000235	4.19E-02	3	Abnormality of the fontanelles or cranial

				sutures
hp	HP:0000234	1.08E-02	22	Abnormality of the head
hp	HP:0000225	1.95E-03	2	Gingival bleeding
hp	HP:0000168	2.69E-02	2	Abnormality of the gingiva
hp	HP:0000157	2.69E-02	3	Abnormality of the tongue
hp	HP:0000152	1.23E-02	22	Abnormality of head or neck
hp	HP:0000144	2.46E-02	2	Decreased fertility
hp	HP:0000140	1.81E-02	3	Abnormality of the menstrual cycle
hp	HP:0000113	1.23E-02	2	Polycystic kidney dysplasia
hp	HP:0000112	2.05E-02	2	Nephropathy
hp	HP:0000107	2.32E-02	3	Renal cyst
hp	HP:0000100	2.05E-02	2	Nephrotic syndrome
hp	HP:0000050	3.00E-02	4	Hypoplastic male external genitalia
hp	HP:0000036	4.52E-02	5	Abnormality of the penis
hp	HP:0000035	4.59E-02	6	Abnormality of the testis
hp	HP:0000014	4.65E-02	2	Abnormality of the bladder
hp	HP:0000007	2.00E-02	22	Autosomal recessive inheritance
hp	HP:0000006	5.00E-02	13	Autosomal dominant inheritance
hp	HP:0000005	1.77E-02	32	Mode of inheritance
hp	HP:0000003	2.92E-02	2	Multicystic kidney dysplasia
BP	GO:0051049	5.00E-02	27	regulation of transport
MF	GO:0004908	7.64E-03	4	interleukin-1 receptor activity