

Supplementary information, Table S11 Functional enrichment categories of PSGs detected by XP-CLR

Term	Term ID	P-value	Gene number	Description
hp	HP:0012232	1.05E-02	2	Shortened QT interval
hp	HP:0001279	2.20E-03	4	Syncope
hp	HP:0001634	3.26E-02	3	Mitral valve prolapse
hp	HP:0002297	1.05E-02	2	Red hair
hp	HP:0100678	2.13E-02	2	Premature skin wrinkling
hp	HP:0002213	2.63E-02	4	Fine hair
hp	HP:0000591	2.63E-02	4	Abnormality of the sclera
hp	HP:0000592	1.63E-02	4	Blue sclerae
hp	HP:0005115	1.03E-02	4	Supraventricular arrhythmia
hp	HP:0001649	1.75E-02	5	Tachycardia
hp	HP:0004755	9.13E-03	4	Supraventricular tachycardia
hp	HP:0001692	9.13E-03	4	Primary atrial arrhythmia
hp	HP:0005110	6.99E-03	4	Atrial fibrillation
hp	HP:0000639	1.09E-02	18	Nystagmus
hp	HP:0010647	1.81E-02	6	Abnormal elasticity of skin
hp	HP:0008067	1.50E-02	5	Abnormally lax or hyperextensible skin
hp	HP:0000977	4.90E-03	3	Soft skin
hp	HP:0002047	3.54E-02	2	Malignant hyperthermia
hp	HP:0000993	6.39E-03	2	Molluscoid pseudotumors
hp	HP:0008056	1.18E-02	10	Aplasia/Hypoplasia affecting the eye
hp	HP:0008062	2.71E-02	5	Aplasia/Hypoplasia affecting the anterior segment of the eye
hp	HP:0008055	4.60E-02	4	Aplasia/Hypoplasia affecting the uvea
hp	HP:0008053	4.28E-02	4	Aplasia/Hypoplasia of the iris
hp	HP:0001788	2.13E-02	2	Premature rupture of membranes
hp	HP:0002445	3.54E-02	2	Tetraplegia
hp	HP:0001250	3.65E-02	23	Seizures
hp	HP:0000963	4.23E-03	5	Thin skin
hp	HP:0004329	2.66E-02	20	Abnormality of the posterior segment of the eye
hp	HP:0001252	5.00E-02	19	Muscular hypotonia
hp	HP:0100738	3.54E-02	2	Abnormal eating behavior
hp	HP:0001103	2.04E-02	5	Abnormality of the macula
hp	HP:0000493	1.55E-02	2	Abnormality of the fovea
hp	HP:0008061	2.77E-03	3	Aplasia/Hypoplasia affecting the retina
hp	HP:0008059	1.97E-03	3	Aplasia/Hypoplasia of the macula
hp	HP:0008060	1.05E-02	2	Aplasia/Hypoplasia of the fovea
hp	HP:0007750	1.05E-02	2	Hypoplasia of the fovea
hp	HP:0001030	4.36E-02	2	Fragile skin

hp	HP:0002085	4.36E-02	2	Occipital encephalocele
ke	KEGG:04144	1.27E-02	7	Endocytosis
ke	KEGG:04020	1.79E-02	6	Calcium signaling pathway
ke	KEGG:04270	6.05E-03	5	Vascular smooth muscle contraction
ke	KEGG:00230	3.49E-02	5	Purine metabolism
ke	KEGG:04916	4.73E-02	3	Melanogenesis
ke	KEGG:00565	2.88E-02	2	Ether lipid metabolism
ke	KEGG:00520	1.45E-02	3	Amino sugar and nucleotide sugar metabolism
ke	KEGG:00650	1.76E-02	2	Butanoate metabolism
ke	KEGG:04060	4.09E-02	5	Cytokine-cytokine receptor interaction
ke	KEGG:04350	2.81E-03	5	TGF-beta signaling pathway
ke	KEGG:00410	1.63E-02	2	beta-Alanine metabolism
ke	KEGG:04260	1.69E-02	3	Cardiac muscle contraction
ke	KEGG:04010	4.69E-02	6	MAPK signaling pathway
ke	KEGG:00280	3.93E-02	2	Valine, leucine and isoleucine degradation
ke	KEGG:04510	3.22E-02	6	Focal adhesion
ke	KEGG:04110	3.52E-02	4	Cell cycle
ke	KEGG:04210	2.89E-02	3	Apoptosis
ke	KEGG:00983	2.16E-02	2	Drug metabolism - other enzymes