Table 13. Genes showing significant signal of postive selection and associated with human disease

Lineage	Ensembl gene ID	Gene name	Gene description *	OMIM ID	Disease description †
Both	ENSG00000141458	NPC1	Niemann-Pick C1 protein precursor.	257220	Niemann-Pick disease, type C
				257220	Niemann-Pick disease, type D
Chimp	ENSG00000001626	CFTR	Cystic fibrosis transmembrane conductance regulator (CFTR) (cAMP- dependent chloride channel) (ATP-binding cassette transporter sub- family C member 7).	602421	Congenital bilateral absence of vas deferens
				602421	Cystic fibrosis
				602421	Sweat chloride elevation without CF
	ENSG00000100312	ACR	Acrosin precursor (EC 3.4.21.10) [Contains: Acrosin light chain; Acrosin heavy chain].	102480	Male infertility due to acrosin deficiency
	ENSG00000100368	CSF2RB	Cytokine receptor common beta chain precursor (GM-CSF/IL-3/IL-5 receptor common beta-chain) (CD131 antigen) (CDw131).	138981	Pulmonary alveolar proteinosis
	ENSG00000101076	HNF4A	Hepatocyte nuclear factor 4-alpha (HNF-4-alpha) (Transcription factor HNF-4) (Transcription factor 14).	600281	MODY, type 1
				600281	Non-insulin-dependent diabetes mellitus
	ENSG00000103449	SALL1	Sal-like protein 1 (Zinc finger protein SALL1) (Spalt-like transcription factor 1) (HSal1).	602218	Townes-Brocks syndrome
	ENSG00000113905	HRG	Histidine-rich glycoprotein precursor (Histidine-proline-rich glycoprotein) (HPRG).	142640	Thrombophilia due to elevated HRG
	ENSG00000135346	CGA	Glycoprotein hormones alpha chain precursor (Anterior pituitary glycoprotein hormones common alpha subunit) (Follitropin alpha chain) (Follicle-stimulating hormone alpha chain) (FSH-alpha) (Lutropin alpha chain) (Luteinizing hormone alpha chain) (LSH-alph	307150	Hypertrichosis, congenital generalized
	ENSG00000135605	TEC	Tyrosine-protein kinase Tec (EC 2.7.1.112).	148500	Tylosis with esophageal cancer
	ENSG00000173230	GOLGB1	Golgin subfamily B member 1 (Giantin) (Macrogolgin) (372 kDa Golgi complex-associated protein) (GCP372).	303800	Colorblindness, deutan
	ENSG00000197912	SPG7	Paraplegin (EC 3.4.24.-) (Spastic paraplegia protein	602783	Spastic paraplegia-7
Human	ENSG00000102805	CLN5	Ceroid-lipofuscinosis neuronal protein 5 (Protein CLN5).	256731	Ceroid-lipofuscinosis, neuronal-5, variant late infantile
	ENSG00000122971	ACADS	Acyl-CoA dehydrogenase, short-chain specific, mitochondrial precursor (EC 1.3.99.2) (SCAD) (Butyryl-CoA dehydrogenase).	201470	Acyl-CoA dehydrogenase, short-chain, deficiency of
	ENSG00000133805	AMPD3	AMP deaminase 3 (EC 3.5.4.6) (AMP deaminase isoform E) (Erythrocyte AMP deaminase).	102772	AMP deaminase deficiency, erythrocytic

ENSG00000138029	HADHB	Trifunctional enzyme beta subunit, mitochondrial precursor (TP-beta) [Includes: 3-ketoacyl-CoA thiolase (EC 2.3.1.16) (Acetyl-CoA acyltransferase) (Beta-ketothiolase)].	143450	Trifunctional protein deficiency, type II
ENSG00000141837	CACNA1A	Voltage-dependent P/Q-type calcium channel alpha-1A subunit (Voltage- gated calcium channel alpha subunit Cav2.1) (Calcium channel, L type, alpha-1 polypeptide isoform 4) (Brain calcium channel I) (BI).	601011 601011 601011 601011	Cerebellar ataxia, pure Episodic ataxia, type 2 Hemiplegic migraine, familial Spinocerebellar ataxia-6
ENSG00000147889	CDKN2A	Cyclin-dependent kinase inhibitor 2A, isoform 4 (p14ARF) (p19ARF).	155600 600160	Malignant melanoma, cutaneous Melanoma
ENSG00000163069	SGCB	Beta-sarcoglycan (Beta-SG) (43 kDa dystrophin-	600900	Muscular dystrophy, limb-girdle, type
ENSG00000165125	TRPV6	Transient receptor potential cation channel subfamily V member 6 (TrpV6) (Epithelial calcium channel 2) (ECaC2) (Calcium transport protein 1) (CaT1) (CaT-like) (CaT-L).	600184	Carnitine acetyltransferase deficiency
ENSG00000165409	TSHR	Thyrotropin receptor precursor (TSH-R) (Thyroid-stimulating hormone receptor).	275200 275200 275200 275200	Graves disease Hyperthroidism, congenital Hypothyroidism, nongoitrous, due to Thyroid adenoma, hyperfunctioning
ENSG00000169738	DCXR	L-xylulose reductase (EC 1.1.1.10) (XR) (Dicarbonyl/L-xylulose reductase) (Kidney dicarbonyl reductase) (kiDCR) (Carbonyl reductase II) (Sperm surface protein P34H).	190685	Down syndrome
ENSG00000180509	KCNE1	Potassium voltage-gated channel subfamily E member 1 (IKs producing slow voltage-gated potassium channel beta subunit Mink) (Minimal potassium channel) (Delayed rectifier potassium channel subunit IsK).	176261	Jervell and Lange-Nielsen syndrome
ENSG00000182372	CLN8	Protein CLN8.	600143	Epilepsy, progressive, with mental retardation
ENSG00000186395	KRT10	Keratin, type I cytoskeletal 10 (Cytokeratin-10) (CK-10) (Keratin-10) (K10).	148080	Epidermolytic hyperkeratosis
ENSG00000188153	COL4A5	Collagen alpha-5(IV) chain precursor	303630 303630	Alport syndrome Leiomyomatosis-nephropathy syndrome

* Gene descriptions downloaded from Ensembl.

† Disease descriptions downloaded from OMIM.