

Supplemental File 6

Common genes subject to hemizygous deletion in overlapping region of Cohort A

Patients 1 and 2 (and seven genes in a region of hemizygous deletion in Patient 2 but not Patient 1)

Accession	Symbol	Name
NM_020639	RIPK4	Receptor-interacting serine-threonine kinase 4
NM_001040424	PRDM15	PR domain containing 15
NM_199050	C2CD2	C2 calcium-dependent domain containing 2
NM_020727	ZNF295	Zinc finger protein 295
NM_173568	UMODL1	Uromodulin-like 1
NM_207627	ABCG1	ATP-binding cassette, sub-family G (WHITE), member 1
NM_003226	TFF3	Trefoil factor 3 (intestinal)
NM_005423	TFF2	Trefoil factor 2 (spasmolytic protein 1)
NM_003225	TFF1	Trefoil factor 1
NM_032404	TMPRSS3	Transmembrane protease, serine 3
NM_018961	UBASH3A	Ubiquitin associated and SH3 domain containing, A
NM_080860	RSPH1	Radial spoke head 1 homolog (Chlamydomonas)
NM_018964	SLC37A1	Solute carrier family 37 (glycerol-3-phosphate transporter), member 1
NM_002606	PDE9A	Hypothetical protein LOC100132338
NM_033661	WDR4	WD repeat domain 4
NM_001001503	NDUFV3	NADH dehydrogenase (ubiquinone) flavoprotein 3, 10kDa
NM_004571	PKNOX1	PBX/knotted 1 homeobox 1
NM_000071	CBS	Cystathionine-beta-synthase
NM_006758	U2AF1	U2 small nuclear RNA auxiliary factor 1
NM_000394	CRYAA	Crystallin, alpha A
NM_173354	SNF1LK	SNF1-like kinase
NM_007031	HSF2BP	Heat shock transcription factor 2 binding protein
NM_015056	RRP1B	Ribosomal RNA processing 1 homolog B (S. cerevisiae)
NM_003681	PDXK	Pyridoxal (pyridoxine, vitamin B6) kinase
NM_000100	CSTB	Cystatin B (stefin B)
NM_003683	RRP1	Ribosomal RNA processing 1 homolog (S. cerevisiae)
NM_020132	AGPAT3	1-acylglycerol-3-phosphate O-acyltransferase 3
NM_003274	TMEM1	Transmembrane protein 1
NM_005049	PWP2	PWP2 periodic tryptophan protein homolog (yeast)
NM_004649	C21orf33	Chromosome 21 open reading frame 33
NM_015259	ICOSLG	Inducible T-cell co-stimulator ligand
NM_013369	DNMT3L	DNA (cytosine-5-)-methyltransferase 3-like
NM_000383	AIRE	Autoimmune regulator
NM_002626	PFKL	Phosphofructokinase, liver

NM_004928	C21orf2	Chromosome 21 open reading frame 2
NM_003307	TRPM2	Transient receptor potential cation channel
NM_030891	LRRC3	Leucine rich repeat containing 3
NM_144991	C21orf29	Chromosome 21 open reading frame 29
NM_198691	KRTAP10-1	Keratin associated protein 10-1
NM_198693	KRTAP10-2	Keratin associated protein 10-2
NM_198696	KRTAP10-3	Keratin associated protein 10-3
NM_198687	KRTAP10-4	Keratin associated protein 10-4
NM_198694	KRTAP10-5	Keratin associated protein 10-5
NM_198688	KRTAP10-6	Keratin associated protein 10-6
NM_198689	KRTAP10-7	Keratin associated protein 10-7
NM_198695	KRTAP10-8	Keratin associated protein 10-8
NM_198690	KRTAP10-9	Keratin associated protein 10-9
NM_181688	KRTAP10-10	Keratin associated protein 10-10
NM_198692	KRTAP10-11	Keratin associated protein 10-11
NM_198698	KRTAP12-4	Keratin associated protein 12-4
NM_198697	KRTAP12-3	Keratin associated protein 12-3
NM_181684	KRTAP12-2	Keratin associated protein 12-2
NM_181686	KRTAP12-1	Keratin associated protein 12-1
NM_198699	KRTAP10-12	Keratin associated protein 10-12
NM_003343	UBE2G2	Ubiquitin-conjugating enzyme E2G 2 (UBC7 homolog, yeast)
NM_006936	SUMO3	SMT3 suppressor of mif two 3 homolog 3 (<i>S. cerevisiae</i>)
NM_004339	PTTG1IP	Pituitary tumor-transforming 1 interacting protein
NM_000211	ITGB2	Integrin, beta 2 (complement component 3 receptor 3 and 4 subunit)
NM_058190	C21orf70	Chromosome 21 open reading frame 70
NM_001112	ADARB1	Adenosine deaminase, RNA-specific, B1 (RED1 homolog rat)
NM_015227	POFUT2	Protein O-fucosyltransferase 2
NM_130445	COL18A1	Collagen, type XVIII, alpha 1
NM_199175	C21orf123	Chromosome 21 open reading frame 123
NM_030582	COL18A1	Collagen, type XVIII, alpha 1
NM_194255	SLC19A1	Solute carrier family 19 (folate transporter), member 1
NM_001130141	PCBP3	Poly(rC) binding protein 3
NM_001848	COL6A1	Collagen, type VI, alpha 1
NM_058175	COL6A2	Collagen, type VI, alpha 2
NM_006657	FTCD	Formiminotransferase cyclodeaminase
NM_032261	C21orf56	Chromosome 21 open reading frame 56
NM_001001438	LSS	Lanosterol synthase (2,3-oxidosqualene-lanosterol cyclase)
NM_003906	MCM3AP	Minichromosome maintenance complex component 3 associated protein
NM_001006114	C21orf57	Chromosome 21 open reading frame 57
NM_058180	C21orf58	Chromosome 21 open reading frame 58
NM_006031	PCNT	Pericentrin
NM_206891	DIP2A	DIP2 disco-interacting protein 2 homolog A (<i>Drosophila</i>)

NM_006272	S100B	S100 calcium binding protein B
NM_001535	PRMT2	Protein arginine methyltransferase 2
NM_012105	BACE2	(Patient 2 only) Beta-site APP-cleaving enzyme 2
NM_182832	PLAC4	(Patient 2 only) Placenta-specific 4
NM_058186	FAM3B	(Patient 2 only) Family with sequence similarity 3, member B
NM_002463	MX2	(Patient 2 only) Myxovirus (influenza virus) resistance 2 (mouse)
NM_002462	MX1	(Patient 2 only) Myxovirus (influenza virus) resistance 1, interferon-inducible protein p78 (mouse)
NM_005656	TMPRSS2	(Patient 2 only) Transmembrane protease, serine 2
NM_020639	RIPK4	(Patient 2 only) Receptor-interacting serine-threonine kinase 4