

APPENDIX A

TABLE OF MANUALLY CURATED HI GENES

Class definition:

1. Severe dominant developmental disorder, smaller critical region, genes disrupted in multiple patients or in a single patient with strong additional evidence.
2. Severe dominant developmental disorder, genes disrupted in a (large) deletion interval in a single patient, with (or without) other evidence.
3. Recessive disorder, or multigenic diseases resulting from compound mutations, or cancer which requires LOF or compound heterozygosity.
4. Evidence failed to support haploinsufficiency.

Gene	Class	Gene	Class	Gene	Class	Gene	Class
ACVRL1	1	AFF3	1	ALX4	1	ANKRD11	1
APC	1	ATM	1	ATP1A2	1	ATP2A2	1
ATP2C1	1	BDNF	1	BMPR2	1	BRCA2	1
BUB1B	1	CAMTA1	1	CBFB1	1	CD2AP	1
CDKN2A	1	CDX2	1	CHD2	1	CHD7	1
CHRNA7	1	CNTNAP2	1	COL1A1	1	COL3A1	1
COL5A1	1	CREEBP	1	CYP11A1	1	DMPK	1

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Gene	Class	Gene	Class	Gene	Class	Gene	Class
DSPP	1	DYRK1A	1	EHMT1	1	ELN	1
ELOVL4	1	EXT2	1	EYA1	1	FBN1	1
FGF10	1	FGF3	1	FGF8	1	FGFR1	1
FGFR2	1	FLG	1	FOXC1	1	FOXE3	1
FOXF1	1	FOXG1	1	FOXL2	1	FOXP2	1
GATA4	1	GDF5	1	GLI3	1	GNB1L	1
GNRH1	1	GPR98	1	GRN	1	GTF2I	1
GTF2IRD1	1	HMGA2	1	HNF1B	1	HOXD13	1
IGF1R	1	INSR	1	IRF6	1	ITPR1	1
JAG1	1	KCNAB2	1	KRIT1	1	KRT14	1
LEMD3	1	LMX1B	1	MBD5	1	MBP	1
MEF2C	1	MSX1	1	MSX2	1	NF1	1
NFIA	1	NFIB	1	NFKB1	1	NKX2-1	1
NKX2-6	1	NOTCH1	1	NR2F2	1	NR5A1	1
NSD1	1	OPA1	1	PAG1	1	PAX2	1
PAX3	1	PAX5	1	PAX6	1	PBX1	1
PHOX2B	1	PITX2	1	PKD1	1	PRKAR1A	1
PRKD2	1	PTCH1	1	PTEN	1	RAI1	1
RALGAPA1	1	ROR2	1	RPS14	1	RPS19	1
RUNX2	1	SALL1	1	SCN1A	1	SERPINA6	1
SGCE	1	SHH	1	SHOX	1	SLC2A1	1
SMAD5	1	SOCS1	1	SOX10	1	SOX2	1
SOX9	1	SRGAP3	1	STK11	1	STXBP1	1
SUMO1	1	TAB2	1	TBX3	1	TBX5	1
TBX6	1	TECTA	1	TIMM23	1	TTF1	1
TWIST1	1	UFD1L	1	WT1	1	ZEB2	1
ABCA3	2	ADAR	2	ANXA7	2	APAF1	2
ARFGAP1	2	ATR	2	ATXN1	2	BAZ1B	2
BLM	2	BMP4	2	BRCA1	2	BUB3	2
C10orf11	2	CASK	2	CDC73	2	CDKL3	2
CDKN1B	2	CDKN1C	2	CELF2	2	CHEK1	2
CHRNA4	2	CNTN4	2	COL6A1	2	COMT	2
COPS3	2	CSH1	2	CTCF	2	CYFIP1	2
DFFB	2	DLL4	2	DMRT1	2	DOCK1	2
DSG1	2	EFNB2	2	EGR1	2	EME1	2

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Gene	Class	Gene	Class	Gene	Class	Gene	Class
ENG	2	ERBB4	2	ETV6	2	FECH	2
FLCN	2	FLII	2	FOXC2	2	FOXL1	2
FOXO3	2	FZD4	2	GATA3	2	GDNF	2
GNAS	2	GPC1	2	GPR35	2	HFE	2
HIC1	2	HIRA	2	HNF1A	2	ID2	2
IGF1	2	IGFBP3	2	ITGB6	2	KCNQ1	2
KCNQ2	2	KHDRBS1	2	KIAA2022	2	KLF4	2
KLF6	2	LETM1	2	MAGOH	2	MAP3K4	2
MC4R	2	MED15	2	MITF	2	MLL	2
MNX1	2	MSH2	2	MSH6	2	MUS81	2
MYCN	2	MYF6	2	MYH9	2	NBN	2
NCF1	2	NF2	2	NFRKB	2	NKX2-5	2
NKX3-1	2	NOG	2	NPAS3	2	NPM1	2
NR2F1	2	NUP98	2	P2RY12	2	PAFAH1B1	2
PAX9	2	PCGF2	2	PDX1	2	PML	2
PRM2	2	QKI	2	RAD50	2	RAD51L1	2
RAE1	2	RB1	2	REEP1	2	RET	2
RNF135	2	RUNX1	2	SALL3	2	SCN2A	2
SCN5A	2	SEMA5A	2	SHANK3	2	SIM1	2
SIX6	2	SLC1A2	2	SLC4A11	2	SMARCB1	2
STK25	2	TACR1	2	TBX1	2	TCOF1	2
TERT	2	TNXB	2	TOB	2	TP53	2
TP73	2	TSC2	2	TUBGCP5	2	WHSC1	2
WNT2B	2	YWHAG	2	ZIC2	2	ADCY9	3
ARFGAP3	3	CADM1	3	CAV1	3	CFH	3
CHN2	3	COL11A1	3	COL1A2	3	CYP2A6	3
DGKD	3	DMRT2	3	DNAJC3	3	EXOC6B	3
FADD	3	FAS	3	FAT1	3	FGFR3	3
FKBP6	3	FOXO1	3	GGCX	3	GPD2	3
GPSM2	3	H2AX	3	IFI44	3	IKZF1	3
IL3RA	3	IRF8	3	KCNRG	3	KLF2	3
LIMK1	3	MAP4K2	3	NGF	3	NR5A2	3
NTNG2	3	PACRG	3	PAPSS2	3	PAX1	3
PINK1	3	PTHLH	3	RHOBTB1	3	RNF139	3
RPS4X	3	SERPIND1	3	SHFM1	3	SLC26A4	3

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Gene	Class	Gene	Class	Gene	Class	Gene	Class
SMAD4	3	SPI1	3	SPINK5	3	SRL	3
SSSCA1	3	SUFU	3	TNS3	3	UTP6	3
VAV3	3	WRD	3	ANGPT2	4	ATP2B2	4
COPS5	4	CRX	4	CTCFL	4	ENOSF1	4
FBXW7	4	FEN1	4	GPR172A	4	GSK3B	4
HAND2	4	HOXA1	4	IGFALS	4	MCL	4
MUTYH	4	MYOC	4	NR3C2	4	P2RY8	4
PARK7	4	PPARG	4	PRG2	4	PRODH	4
PROKR2	4	PXMP2	4	RELA	4	RORA	4
SH3PXD2A	4	SMN2	4	SMS	4	SOCS2	4
SOX18	4	SPAST	4	STAT5A	4	STK19	4
SUB1	4	TAS2R38	4	TCF7L2	4	TGIF1	4
TRPS1	4	XRCC5	4				