

GT HapScreen® Kits

Name Of Kit	Disease	Gene	Chr. No.
GT HBA HapScreen®	Alpha Thalassemia	HBA	16
GT HBB HapScreen®	Beta Thalassemia	HBB	11
GT F8 HapScreen®	Hemophilia A	F8	X
GT F9 HapScreen®	Hemophilia B	F9	X
GT F5 HapScreen®	Coagulation Factor 5 Deficiency	F5	1
GT F7 HapScreen®	Coagulation Factor 7 Deficiency	F7	13
GT F11 HapScreen®	Coagulation Factor 11 Deficiency	F11	4
GT F13A HapScreen®	Coagulation Factor 13A Deficiency	F13A	6
GT F13B HapScreen®	Coagulation Factor 13 B Deficiency	F13B	1
GT vWF HapScreen®	von Willebrand	vWF	12
GT MCFD2 HapScreen®	Combined Deficiency F5-F8	MCFD2	2
GT LMAN1 HapScreen®	Combined Deficiency F5-F8	LMAN1	18
GT ITGA2B & ITGB3 HapScreen®	Glansmann	ITGA2B & ITGB3	17
GT PAH HapScreen®	Phenylketonuria, PKU	PAH	12
GT PTS HapScreen®	BH4 Deficiency Phenylketonuria, BH4 PKU	PTS	11
GT QDPR HapScreen®	BH4 Deficiency Phenylketonuria, BH4 PKU	QDPR	4
GT GCH HapScreen®	BH4 Deficiency Phenylketonuria, BH4 PKU	GCH	14
GT PCBD HapScreen®	BH4 Deficiency Phenylketonuria, BH4 PKU	PCBD	10
GT SPR HapScreen®	BH4 Deficiency Phenylketonuria, BH4 PKU	SPR	2
GT Dystrophin HapScreen®	Duchene Muscular Dystrophy and Becker Muscular Dystrophy, DMD & BMD	Dystrophin	X
GT SGCA HapScreen®	Limb Girdle Muscular Dystrophy, LGMD	SGCA	17
GT SGCG HapScreen®	Limb Girdle Muscular Dystrophy, LGMD	SGCG	13
GT DYSF HapScreen®	Limb Girdle Muscular Dystrophy, LGMD	DYSF	2
GT CAPN3 HapScreen®	Limb Girdle Muscular Dystrophy, LGMD	CAPN3	15
GT SGCD HapScreen®	Limb Girdle Muscular Dystrophy, LGMD	SGCD	5
GT SGCB HapScreen®	Limb Girdle Muscular Dystrophy, LGMD	SGCB	4
GT LAMB3 HapScreen®	Junctional Epidermolysis Bullosa	LAMB3	1
GT COL7A1 HapScreen®	Dystrophic Epidermolysis Bullosa, DEB	COL7A1	3
GT FERMT1 HapScreen®	Kindler	FERMT1	1
GT TGM1 HapScreen®	Ictiosis Lamelar	TGM1	14
GT BRCA1 HapScreen®	Breast Cancer	BRCA1	17
GT BRCA2 HapScreen®	Breast Cancer	BRCA2	13

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GT CYP21A2 HapScreen®	Congenital Adrenal Hyperplasia, CAH	CYP21A2	6
GT AR HapScreen®	Androgen Insensitivity Syndrome	AR	X
GT ASS1 HapScreen®	Citrullinemia	ASS1	9
GT GALK1 HapScreen®	Galactosemia	GALK1	17
GT GALE HapScreen®	Galactosemia	GALE	1
GT GALT HapScreen®	Galactosemia	GALT	9
GT BCKDHA HapScreen®	Maple Syrup Urine Disease, MSUD	BCKDHA	19
GT BCKDHB HapScreen®	Maple Syrup Urine Disease, MSUD	BCKDHB	6
GT DLD HapScreen®	Maple Syrup Urine Disease, MSUD	DLD	17
GT DBT HapScreen®	Maple Syrup Urine Disease, MSUD	DBT	1
GT MMAA HapScreen®	Methyl Malonic Aciduria, MMA	MMAA	4
GT MMAB HapScreen®	Methyl Malonic Aciduria, MMA	MMAB	12
GT MUT HapScreen®	Methyl Malonic Aciduria, MMA	MUT	6
GT SGCH HapScreen®	Mucopolysaccharidosis, MPS	SGCH	17
GT IDUA HapScreen®	Mucopolysaccharidosis, MPS	IDUA	9
GT NAGLU HapScreen®	Mucopolysaccharidosis, MPS	NAGLU	9
GT SMPD HapScreen®	Neimann Pick Type A & B	SMPD	11
GT GAA HapScreen®	Pompe	GAA	17
GT HEXB HapScreen®	Sandhoff Syndrome	HEXB	5
GT FAH HapScreen®	Tyrosinemia	FAH	15
GT TAT HapScreen®	Tyrosinemia	TAT	16
GT SLC37A4 HapScreen®	von Gierke	SLC37A4	11
GT WFS1 HapScreen®	Walfram Syndrome	WFS1	4
GT CSF1R HapScreen®	Adult-Onset Leukoencephalopathy with Axonal Spheroids and Pigmented Glia	CSF1R	5
GT TYR HapScreen®	Albinism	TYR	9
GT USH2AR HapScreen®	Asher Syndrome	USH2AR	5
GT CFTR HapScreen®	Cystic Fibrosis, CF	CFTR	7
GT GJB2 HapScreen®	Heridity Deafness	GJB2	13
GT FANCG HapScreen®	Fanconi Anemia	FANCG	9
GT STX11 HapScreen®	Familial Haemophagocytic Lymphohistiocytosis 4, FHL4	STX11	6
GT MEFV HapScreen®	Familiar Mediteranean Fever, FMF	MEFV	16