DISEASE	NO. OF LINES	INHERITANCE	MUTATION	AFFECTED TISSUE
β-thalassemia	1	AR	homozygote N37/A in the HBB	red blood cells
Gaucher	1	AR	compound heterozygote 84 GG insertion/R496H in β-glucocerebrosidase	nerve, bone marrow, liver, spleen
Pompe	1	AR	R819P G>C in GAA	muscle, heart, liver
CIPA	1	AR	1926 ins T in TrkA	nerve
Cystic fibrosis	1	AR	homozygote for W1282X in CFTR	pancreas, intestinal glands , bronchial glands , sweat glands , sertoli cells
Fanconi C	1	AR	IVS4 +4A>T mutation in the Fanc C	blood
Retinoblastoma	4	AD	C1572insAA in Rb	retina
BRCA1	2	AD	5382 insC in BRCA1	breast and ovary
BRCA2	2	AD	6174del T in BRCA2	breast and ovary
HNPCC	1	AD	V51D mutation	colon
Von Hippel Lindau	1	AD	R64P in VHL	tumors and cysts in multiple organs
Neurofibromatosis	4	AD	c.4269+1G>C in NF	nerve and skin
Paroxysmal Kinesigenic Dyskinesia (together with DMD)	1	AD	c.649dup C in the PRRT2 gene	nerve
Polycystic Kidney	1	AD	mutation 12178C>T in the PKD1 gene	kidney
Branchio-Oto-Renal syndrome	2	AD	433del G in exon 5 of EYA1	kidney, ear, nerve, liver, spleen
Hypohidrotic ectodermal dysplasia	2	AD	D50N in GJB2	hair-nail, skin, nerve
Marfan	1	AD	Q1813X in the FBN1	skeletal, cardiac, connective tissues, lens

DISEASE	NO. OF LINES	INHERITANCE	MUTATION	AFFECTED TISSUE
SMA	2	AR	deletion in exon 7 in SMN1	nerve
Emery Dreifuss	1	AD	4bp deletion in exon 6 of the Emerin	skeletal and cardiac muscles
Osteogenesis imperfecta	1	AD	c.1081C>T mutation in the COL1A1gene	bone
Dyskeratosis Congenital	3	AD	mutation in the catalytic site of TERT	bone marrow failure
Congenital central hypoventilation (CCHS)	1	AD	Poly A expansion in the PHOX2B	nerve
Huntington	2	AD	CAG expansion in Huntingtin	nerve
Myotonic dystrophy type 1	16	AD	CTG expansion in DMPK	skeletal and smooth muscles, cardiac conduction, nerve, endocrine glands, eye lens
C9/ALS-FTD	2	AD	G_4C_2 expansion in C9orf72	motor neurons
Fragile X syndrome/FXTAS	19	X-linked	CGG expansion in FMR1	nerve
SBMA (Kennedy Disease)	1	X-linked	>40 CAG repeats in the Androgen receptor	muscle and breast
Hunter	1	X-linked	exon 4-7 del in the IDS	nerve
Charcot Marie Tooth	1	X-linked	C223T in the GJB1	nerve
Duchenne Muscular Dystrophy	3	X-linked	deletion/duplication	muscle