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 FancC	blood
Retinoblastoma	4	AD	C1572insAA in Rb	retina
BRCA1	1	AD	5382 insC in BRCA1	breast and ovary
BRCA2	2	AD	carrier of 6174del T in BRCA2	breast and ovary
HNPCC	1	AD	V51D mutation	colon
Von Hippel Lindau	1	AD	VHL	multisystem
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
SMA	2	AD	carriers of a deletion in exon 7 in SMN1	nerve
Emery Dreifuss	1	AD	4bp deletion in exon 6 of the Emerin	skeletal and cardiac muscles

DISEASE	NO. OF LINES	INHERITANCE	MUTATION
β-thalassemia	1	AR	homozygote N3
Gaucher	1	AR	compound hete insertion/R496H glucocerebrosid
Pompe	1	AR	R819P G>C in G
CIPA	1	AR	1926 ins T in Trl
Cystic fibrosis	1	AR	homozygote for CFTR
Fanconi C	1	AR	IVS4 +4A>T mut FancC
Retinoblastoma	4	AD	C1572insAA in F
BRCA1	1	AD	5382 insC in BR
BRCA2	2	AD	carrier of 6174
HNPCC	1	AD	V51D mutation
Von Hippel Lindau	1	AD	VHL
Branchio-Oto-Renal syndrome	2	AD	433del G in exo
Hypohidrotic ectodermal dysplasia	2	AD	D50N in GJB2
Marfan	1	AD	Q1813X in the

DISEASE	NO. OF LINES	INHERITANCE	MUTATION
SMA	2	AD	carriers of a deletion in e in SMN1
Emery Dreifuss	1	AD	4bp deletion in exon 6 of Emerin
Polycystic kidney	1	AD	carrier of deletion c.8433_8438delinsGCCC i PCKD1
Dyskeratosis Congenital	3	AD	mutation in the catalytic s TERT
Myotonic dystrophy type 1	15	AD	CTG expansion in DMPK
C9/ALS-FTD	2	AD	G_4C_2 expansion in C9orf7
Fragile X syndrome/FXTAS	15	X-linked	CGG expansion in FMR1
Hunter	1	X-linked	exon 4-7 del in the IDS
Charcot Marie Tooth	1	X-linked	C223T in the GJB1
SBMA (Kennedy Disease)	1	X-linked	>40 CAG repeats in the Androgen receptor
Duchenne Muscular Dystrophy	1	X-linked	deletion
Alport	1	X-linked	c.4323delT at the gene COL4A5
TOTAL	**67**		