

Examples of available immortalized human cells*

Pathology/ Gene	Mutation	Cell ID	Type of cells	Muscle-Tissu	Age	Sex
Control	-	KM1421	myoblasts	Paravertebral	13 years	F
Control	-	AB918	MyoD-Fibroblasts	Skin	14 years	F
Control	-	AB1190	myoblasts	Paravertebral	16 years	M
Control	-	AB1191	MyoD-Fibroblasts	Skin	16 years	M
Control	-	AB943	MyoD-Fibroblasts	Skin	16 years	M
Control	-	AB1035	MyoD-Fibroblasts	Skin	16 years	M
Control	-	AB1190FAPs	FAPs	Paravertebral	16 years	M
Control	-	AB1167	myoblasts	Fascia lata	20 years	M
Control-GFP	-	AB1167-GFP	myoblasts	Fascia lata	20 years	M
Control	-	AB1436	myoblasts	Fascia lata	33 years	F
Control	-	AB1079	myoblasts	Quadriceps	38 years	M
Control	-	AB1079FAPs	FAPs	Quadriceps	38 years	M
Control	-	AB678	myoblasts	Quadriceps	53 years	F
Control-GFP	-	AB678-GFP	myoblasts	Quadriceps	53 years	F
Control	-	KM670	myoblasts	Quadriceps	83 years	M
Col6A1	c.958-8_961del /p. Gly320-Lys322del	KM1154	myoblasts	Paravertebral	10 years	M
Col6A1	HTZ deep intronic (intron 11) with néoexon 72bp c.930+189 C>T intron 11	KM1310	myoblasts	Dorsal	13 years	M

Col6A1	HTZ deep intronic (intron 11) with néoexon 72bp c.930+189 C>T intron 11	KM1310FAPs	FAPs	Dorsal	13 years	M
DMD	del 8-16	AB1239FAPs	FAPs	Paravertebral	14 years	M
DMD	Duplication exon 10-11	KM1316	myoblasts	Paravertebral	14 years	M
DMD	c35456G>T (p.Glu1186X) exon 26	AB1324	myoblasts	Paravertebral	14 years	M
DMD	Del 45	AB1323	myoblasts	Paravertebral	14 years	M
DMD	Del 45	AB1323FAPs	FAPs	Paravertebral	14 years	M
DMD	Del 45-50	KM1315	myoblasts	Paravertebral	14 years	M
DMD	Del 45-50	AB1064	MyoD-Fibroblasts	Skin	17 years	M
DMD	Del 45-52	AB1071	myoblasts	Paravertebral	13 years	M
DMD	Del 45-52	AB1072	MyoD-Fibroblasts	Skin	13 years	M
DMD	Del 45-52	AB1071FAPs	FAPs	Paravertebral	13 years	M
DMD	Del 48-50	AB1098	myoblasts	Paravertebral	14 years	M
DMD	Del 48-50	AB1099	MyoD-Fibroblasts	Skin	14 years	M
DMD	Del 51-54	KM1395	myoblasts	Paravertebral	13 years	M
DMD	Del 51	AB1245	myoblasts	Paravertebral	14 years	M
DMD	Del 51	AB1244	Fibroblasts	Skin	14 years	M
DMD	Del 51-54	KM1395	myoblasts	Paravertebral	13 years	M
DMD	Del 51-55	AB1131	MyoD-Fibroblasts	Skin	13 years	M
DMD	Del 52	KM1328	myoblasts	Paravertebral	16 years	M

DMD	Stop exon 59: c.8713C>T, p.Arg2905X	AB1023	myoblasts	Quadriceps	11 years	M
DMD	Stop exon 59: c.8713C>T, p.Arg2905X	AB1023FAPs	FAPs	Quadriceps	11 years	M
DMD	Stop exon 59: c.8713C>T, p.Arg2905X	AB1024	Fibroblasts	Skin	11 years	M
DMD	Mutation in intron 70 c.IVS70+1 G>A	KM1329	myoblasts	Paravertebral	17 years	M
DM1	2600CTG	ASA308	myoblasts	Gastrocnemius	11 years	F
FSHD	6,3 repeat D4Z4	AB1080	myoblasts	Paravertebral	26 years	M
FSHD	6,3 repeat D4Z4	AB1080FAPs	FAPs	Paravertebral	26 years	M
FSHD	2 repeat D4Z4	KM1263	myoblasts	Rhomboide	21 years	M
LGMD2C	GSG: HMZ c.del525T	KM1090	myoblasts	paravertebral	16 years	F
LGMD2I	FKRP HTZ : c.899T>C / dup 22 bases in c. 798 819	KM977	myoblasts	Paravertebral	17 years	M
MDC1A	LAMA2 HTZ: c.1793_1795del (exon13) & c.3294delG(exon23)	AB1481	myoblasts	Paravertebral	12 years	F
MDC1A	LAMA2 HTZ: c.1793_1795del (exon13) & c.3294delG(exon23)	AB1481FAPs	FAPs	Paravertebral	12 years	F
Myasthenia gravis	Anti AchR	KM1422	myoblasts	Long dorsal	59 years	M
OPMD	(GCG)10 / 15	ABo1440	myoblasts	Cricopharyngeal	52 years	M
OPMD	(GCG)10/13	KM1445	myoblasts	Cricopharyngeal	77 years	M
OPMD	(GCG)10/13	CT1256O	myoblasts	Cricopharyngeal	63 years	M
OPMD	(GCG)10/13	CT1267	myoblasts	Cricopharyngeal	77 years	M
OPMD	(GCG)10/13	AO1311	myoblasts	Cricopharyngeal	63 years	F
Pompe disease	GSD2 HTZ C.1-45T>G in intron 1/ del exon18	KM1269	myoblasts	Tenseur Fascia Lata	53 years	F

SEPN1-related myopathy	SEPN1	AB1480	myoblasts	Paravertebral	19 years	F
SMA-II	0 SMN1- 3 SMN2	KM1150	myoblasts	Paravertebral	7 years	F
SMA-II		AB1141	myoblasts	Lumbar	13 years	F
Myotubular myopathy	MTM1 hemizygous mutation c.205>T exon4	KM1288	myoblasts	Deltoïde	3 years	M

Other cell lines are available on request. Please contact anne.bigot@sorbonne-universite.fr & vincent.mouly@sorbonne-universite.fr