

## Molecular Diagnostics Portfolio Biodonostia Health Research Institute

Service	Study	Genes	Technique	Time
Cardiology	Long QT Syndrome	KCNQ1, KCNH2, KCNE1, KCNE2, SCN5A	NGS	2 months
	Long QT Syndrome + Short QT Syndrome	KCNQ1, KCNH2, SCN5A, KCNE1, KCNE2, ANK2, KCNJ2, CACNA1C, CAV3, SCN4B, AKAP9, SNTA1, CACNB2, CACNA2D1	NGS	3 months
Neurology	Ataxia / Migraine	CACNA1, KCNA1, CACNB4, SLC1A3	NGS	2 months
	Apert syndrome	FGFR2 (2 point mutations: Ser252Trp, Pro253Arg)	Capillary Sequencing	15 days
	Hypokalemic periodic paralysis	CACNA1S (5 point mutations: p.Arg528His, p.Arg528Gly, p.Arg897Ser, p.Arg1239His y p.Arg1239Gly)	Capillary Sequencing	1 month
	Facioscapulohumeral muscular dystrophy type 1 (FSHD1) <b>(1)</b> <i>* Spanish Reference Center</i>	D4Z4 region, chromosome 4	Southern blot (Linear Gel Electrophoresis -LGE-)	2 months
	Facioscapulohumeral muscular dystrophy type 1 (FSHD1) <b>(2)</b> <i>* Spanish Reference Center</i>	D4Z4 region, chromosome 4	Southern blot (Pulsed Field Gel Electrophoresis -PFGE-)	2 months
	Facioscapulohumeral muscular dystrophy type 1 (FSHD1)	SSLP haplotype (chromosomes 4 and 10 alleles)	PCR y fragment analysis	2-3 weeks
	Facioscapulohumeral muscular dystrophy type 1 (FSHD1): Prenatal <i>* Spanish Reference Center</i>	D4Z4 region, chromosome 4	Southern blot (Linear Gel Electrophoresis -LGE-)	2-3 weeks
	Facioscapulohumeral muscular dystrophy type 1 (FSHD1): Prenatal , indirect analysis made using markers	11 markers in the proximal region to D4Z4 (chromosome 4)	PCR y fragment analysis	2 weeks
	Facioscapulohumeral muscular dystrophy type 2 (FSHD2) <i>* Spanish Reference Center</i>	Region D4Z4 hypomethylation (chromosome 4)	Southern blot (Linear Gel Electrophoresis -LGE-)	2-3 months
	Facioscapulohumeral muscular dystrophy type 2 (FSHD2) <i>* Spanish Reference Center</i>	SMCHD1	NGS	2 months
	Limb girdle muscle dystrophy type 2A (calpain) <i>* Spanish Reference Center</i>	CAPN3 *	Capillary Sequencing (RNA and DNA study)	1-2 months
	Limb girdle muscular dystrophy type 2I	FKRP	Capillary Sequencing	1 month

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Neurology	Scapuloperoneal myopathy linked to the X chromosome	FHL1	Capillary Sequencing	1 month
	Recessive limb-girdle muscular dystrophy	ANO5, CANP3, DYSF, FKRP, GAA, LAMP2, SGCG, SGCA, SGCB, SGCD, TCAP, TRIM32, DAG1, FKTN, POMGNT1, POMT1, POMT2	NGS	3 months
	Dominant dystrophies	VCP, CAV3, LMNA, MYOT, DNAJB6	NGS	2 months
	Congenital dystrophies / myopathies 1	ACTA1, CFL2, CNTN1, DNM2, ITGA7, KBTBD13, LAMA2, MYH7, NEB, TNNT1, TPM2, TPM3, TRIM32, GAA, DPAGT1, MTM1	NGS	2 months
	Congenital dystrophies / myopathies 2+3	FKRP, COL6A1, COL6A2, COL6A3, FHL1, FKTN, LARGE, POMGNT1, POMT1, POMT2, SEPN1	NGS	2 months
	Distal myopathy	MYH7, GNE, MATR3, LDB3 (ZASP), NEB, DES, CRYAB, FLNC, BAG3, ACTA1, MYH2, MYH3, VCP, DYSF, MYOT, KLHL9, CAV3, DNM2, ANO5	NGS	3 months
	Channelopathies	CLCN1, SCN4A, CACNA1S, CACNA1A, KCNE3, KCNA1, KCNJ18	NGS	2 months
	Parkinson's disease	TAU, ATP13A2, DJ1, EIF4G1, FBXO7, GBA, GIGYF2, LRRK2, parkin, PINK1, PLA2G6, SNCA, UCHL1, VPS35, HTRA2, DNAJC6, APOE, SYNJ1	NGS	2 months
	Frontotemporal dementia - ALS	APOE, SORL1, PSEN1, PSEN2, APP, TAU, PGRN, VCP, c9orf72, FUS, TARDBP, TMEM106B, ALS2, PFN1, VAPB, SOD1, OPTN, SQSTM1, NEFH, FIG1, FIG4, ANG, SETX, ATX2, UBQLN2, TAF15	NGS	3 months
	Frontotemporal dementia - ALS	C9orf72 expansion **	PCR and fragment analysis	1 month
	Central core myopathies	RYR1	NGS	2 months
	Frontotemporal dementia (Progranulin)	GRN	Capillary Sequencing	1 month
	Prion protein associated dementia	PRNP	Capillary Sequencing	1 month
	Valosin (IBMPFTD syndrome: Inclusion Body Myopathy –IBM-, Paget disease of bone –PDB- and Frontotemporal Dementia –FTD–)	VCP	Capillary Sequencing	1 month
	Parkinson's disease (Dardarin)	LRRK2 (2 one-off mutations: R1441G, G2019S)	Capillary Sequencing	15 days
	Autosomal dominant lateral temporal epilepsy	LG1	Capillary Sequencing	1 month

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Neurology / Ophthalmology	MERRF	MTTL1 (7512, 8344, 8356, 8363, 8296 mutations)	Capillary Sequencing	1 month
	MELAS	MTTL1 (3243, 3256, 3252, 3093, 3244, 3258, 3271, 3291 mutations)	Capillary Sequencing	1 month
	Nonsyndromic mitochondrial sensorineural deafness	A1555G mutation	Capillary Sequencing	15 days
Neurology / Ophthalmology	Leber Hereditary Optic Neuropathy (LHON)	Primary mutations: 11778, 14484, 3460 + Intermediate mutations: 5244, 15257 + Secondary mutations: 3394, 4160, 4216, 4917, 7444, 9438, 13708, 13730, 14459	Capillary Sequencing	1 month
Pediatric	CHARGE syndrome	CHD7	NGS	2 months
	Cornelia de Lange syndrome	NIPBL, SMC1A, SMC3	NGS	2 months
	Crouzon syndrome	FGFR2	Capillary Sequencing	1 month
	Mitochondrial encephalopathy	POLG	NGS	1 month
	Noonan syndrome	PTPN11, RAF1, KRAS, SOS1	NGS	2 months
Miscellanea	Periodic fever syndrome	TNFRSF1A, MEFV, MVK, SAA1, ELANE, PSTPIP1, LPIN2, NLRP3	NGS	2 months
	Marfan syndrome	FBN1	NGS	2 months
	Stickler syndrome	COL11A1, COL2A1, COL11A2, COL9A1, COL9A2	NGS	2 months
	Breast and ovarian cancer	BRCA1 and BRCA2	NGS	2 months
	Mitochondrial diseases	Complete mitochondrial DNA	NGS	1 month

(1) Routine diagnosis to estimate the size of the gene deletion fragment

Individualized gene studies through **Capillary Sequencing** (**SANGER** sequencing)

(2) Complementary diagnosis made to confirm mosaic or reordering cases between chromosome 4 and 10 (hybrid fragments)

Multiple gene linked to the disease simultaneous study (genetic study panels) through massive sequencing techniques (**NGS**)

\* RNA and DNA study

\*\* Request budget for more than a sample