"MAURO BASCHIROTTO" INSTITUTE FOR RARE DISEASES
MEDICAL GENETICS UNIT
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Contact:

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GENETIC TESTS

Disease Name	Locus	Gene	Method
Adrenoleukodystrophy (ABCD1)	Xq28	ABCD1	Seq. gDNA
Adrenoleukodystrophy (ABCD1)	Xq28	ABCD1	MLPA
Alpha-1 Antitrypsin Deficiency (SERPINA1)	14q32.1	SERPINA1	Seq. gDNA
Alzheimer Disease Type 1 (APP)	19q13.2	APP	Seq. gDNA
Alzheimer Disease Type 2 Linked To Apoe (APOE)	19q13.2	APOE	Seq. hot-spot gDNA
Alzheimer Disease Type 3 (PSEN1)	14q24.3	PSEN1	Seq. gDNA
Alzheimer Disease Type 4 (PSEN2)	1q31-q42	PSEN2	Seq. gDNA
Amyotrophic Lateral Sclerosis 1 (SOD1)	21q22.1	SOD1	Seq. gDNA
Amyotrophic Lateral Sclerosis 4, Juvenile (SETX)	9q34.13	SETX	Seq. gDNA
Amyotrophic Lateral Sclerosis With Frontotemporal Dementia 1 (C9ORF72)	9p21.1	C9ORF72	STR
Amyotrophic Lateral Sclerosis With Frontotemporal Dementia 1 (C9ORF72)	9p21.1	C9ORF72	RP-PCR
Angelman Syndrome (UBE3A)	15q11-q13	UBE3A	Det. UPD
Angelman Syndrome (UBE3A)	15q11-q13	UBE3A	MS-PCR
Angelman Syndrome (UBE3A)	15q11-q13	UBE3A	MS-MLPA
Angelman Syndrome (UBE3A)	15q11-q13	UBE3A	Seq. gDNA
Ataxia With Oculomotor Apraxia Type 2 (SETX)	9q34	SETX	Seq. gDNA
Ataxia, Early-Onset, With Oculomotor Apraxia And Hypoalbuminemia (APTX)	9p13.3	APTX	Seq. gDNA
Autoimmune Polyendocrine Syndrome Type 1 (AIRE)	21q22.3	AIRE	Seq. gDNA
Basal Cell Nevus Syndrome (PTCH1)	9q22.3	PTCH1	Seq. gDNA
Berardinelli-Seip Congenital Lipodystrophy Type 1 (BSCL2)	11q13	BSCL2	Seq. gDNA
Brugada Syndrome (KCNE3)	11q13-q14	KCNE3	Seq. gDNA
Charcot - Marie - Tooth Disease, X-Linked, 1 (GJB1)	Xq13.1	GJB1	Seq. gDNA
Charcot-Marie-Tooth Disease, Demyelinating, Type 1a (PMP22)	17p11.2	PMP22	MLPA
Chorea, Benign Hereditary (NKX2-1)	14q13	NKX2-1	Seq. gDNA

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Disease Name	Locus	Gene	Method
Chorea, Benign Hereditary (NKX2-1)	14q13	NKX2-1	MLPA
Corneal Dystrophy, Avellino Type (TGFBI)	5q31	TGFBI	Seq. hot-spot gDNA
Corneal Dystrophy, Groenouw Type I (TGFBI)	5q31	TGFBI	Seq. hot-spot gDNA
Corneal Dystrophy, Reis - Bucklers Type (TGFBI)	5q31	TGFBI	Seq. hot-spot gDNA
Corneal Dystrophy, Thiel - Behnke Type (TGFBI)	5q31	TGFBI	Seq. hot-spot gDNA
Creutzfeldt - Jakob Disease (PRNP)	20pter-p12	PRNP	Seq. gDNA
Creutzfeldt-Jakob Disease (PRNP)	20pter-p12	PRNP	Seq. gDNA
Cystic Fibrosis (CFTR)	7q31.2	CFTR	Reverse dot-Blot
Cystic Fibrosis (CFTR)	7q31.2	CFTR	Seq. gDNA
Darier - White Disease (ATP2A2)	12q23-q24.1	ATP2A2	Seq. gDNA
Deafness, Aminoglycoside-Induced (MTRNR1)	mtDNA	MTRNR1	Seq. hot-spot mtDNA
Deafness, Autosomal Recessive 1A (GJB2)	13q11-q12	GJB2	Seq. gDNA
Deafness, Autosomal Recessive 1B (GJB6)	13q11-q12	GJB6	AS-PCR
Deafness-Dystonia-Optic Neuronopathy Syndrome (TIMM8A)	Xq22	TIMM8A	Seq. gDNA
Dentatorubral-Pallidoluysian Atrophy (ATN1)	12p13.31	ATN1	STR
Dentatorubral-Pallidoluysian Atrophy (ATN1)	12p13.31	ATN1	RP-PCR
Diabetes Insipidus, Nephrogenic, X-Linked (AVPR2)	Xq28	AVPR2	Seq. gDNA
Epileptic Encephalopathy, Early Infantile, 2 (CDKL5)	Xp22	CDKL5	Seq. gDNA
Farber Lipogranulomatosis (ASAH1)	8p22-p21.3	ASAH1	Seq. gDNA
Fatal Familial Insomnia (PRNP)	20pter-p12	PRNP	Seq. gDNA
Friedreich Ataxia 1 (FXN)	9q13, 9p23-p11	FXN	STR
Friedreich Ataxia 1 (FXN)	9q13, 9p23-p11	FXN	RP-PCR
Friedreich Ataxia 1 (FXN)	9q13, 9p23-p11	FXN	Seq. gDNA
Friedreich Ataxia 1 (FXN)	9q13, 9p23-p11	FXN	Long Range PCR

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Disease Name	Locus	Gene	Method
Frontotemporal Dementia (GRN)	17q21.1,14q24.3	GRN	Seq. gDNA
Glycogen Storage Disease Type V (PYGM)	11q13	PYGM	Seq. gDNA
Gout, Hprt-Related (HPRT1)	Xq26-q27.2	HPRT1	qPCR
Gout, Hprt-Related (HPRT1)	Xq26-q27.2	HPRT1	Seq. cDNA
Gout, Hprt-Related (HPRT1)	Xq26-q27.2	HPRT1	Seq. gDNA
Hemochromatosis (HFE)	6p21.3, 20p12	HFE	Seq. gDNA
Hemochromatosis (HFE)	6p21.3, 20p12	HFE	Seq. hot-spot gDNA
Hemochromatosis, Juvenile (HAMP)	19q13	HAMP	Seq. gDNA
Hemochromatosis, Juvenile (HFE2)	19q13, 1q21	HFE2	Seq. gDNA
Hemochromatosis, Type 3 (TFR2)	7q22	TFR2	Seq. gDNA
Hemochromatosis, Type 4 (SLC40A1)	2q32	SLC40A1	Seq. gDNA
Hepatic Venoocclusive Disease With Immunodeficiency (SP110)	2q37.1	SP110	Seq. hot-spot gDNA
Hereditary Neuropathy With Liability To Pressure Palsies (PMP22)	17p11.2	PMP22	MLPA
Homocysteinemia (CBS)	21q22.3	CBS	Seq. gDNA
Homocystinuria Due To Deficiency Of N(5,10)-Methylenetetrahydrofolate Reductas	1p36.3	MTHFR	Seq. gDNA
Homocystinuria Due To Deficiency Of N(5,10)-Methylenetetrahydrofolate Reductas	1p36.3	MTHFR	Seq. hot-spot gDNA
Huntington Disease (HTT)	4p16.3	HTT	RP-PCR
Huntington Disease (HTT)	4p16.3	HTT	STR
Huntington Disease-Like 2 (JPH3)	16q24.3	JPH3	STR
Hyperferritinemia - Cataract Syndrome (FTL)	19q13.3-q13.4	FTL	Seq. gDNA
Hyperimmunoglobinemia D with recurrent fever (MVK)	12q24	MVK	Seq. gDNA
Hyperkalemic Periodic Paralysis Type 1 (SCN4A)	17q23.1-q25.3	SCN4A	Seq. gDNA
Hypokalemic Periodic Paralysis (KCNE3)	11q13-q14	KCNE3	Seq. gDNA
Hypokalemic Periodic Paralysis Type 1 (CACNA1S)	1q32	CACNA1S	Seq. hot-spot gDNA

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Disease Name	Locus	Gene	Method
Hypokalemic Periodic Paralysis Type 1 (CACNA1S)	1q32	CACNA1S	Seq. gDNA
Hypokalemic Periodic Paralysis Type 2 (SCN4A)	17q23.1-q25.3	SCN4A	Seq. hot-spot gDNA
Krabbe Disease (GALC)	14q31	GALC	PCR per Delezione
Krabbe Disease (GALC)	14q31	GALC	Seq. gDNA
Krabbe Disease (GALC)	14q31	GALC	Seq. hot-spot gDNA
Leber Optic Atrophy (MTND1-4-5-6)	mtDNA	MTND1-4-5-6	Seq.mtDNA
Lesch-Nyhan Syndrome (HPRT1)	Xq26-q27.2	HPRT1	qPCR
Lesch-Nyhan Syndrome (HPRT1)	Xq26-q27.2	HPRT1	Seq. cDNA
Lesch-Nyhan Syndrome (HPRT1)	Xq26-q27.2	HPRT1	Seq. gDNA
Leukodystrophy type 2, Hypomyelinating (GJC2)	1q42.13	GJC2	Seq. gDNA
Li-Fraumeni Syndrome 1 (TP53)	17p13.1	TP53	Seq. gDNA
Li-Fraumeni Syndrome 2 (CHEK2)	22q12.1	CHEK2	Seq. gDNA
Lymphedema-Distichiasis Syndrome (FOXC2)	16q24.3	FOXC2	Seq. gDNA
Malignant Hyperthermia, Susceptibility To, 1 (RYR1)	19q13.1	RYR1	Seq. cDNA
Malignant Hyperthermia, Susceptibility To, 1 (RYR1)	19q13.1	RYR1	Seq. hot-spot gDNA
Malignant Hyperthermia, Susceptibility To, 1 (RYR1)	19q13.1	RYR1	Seq. hot-spot gDNA
Malignant Hyperthermia, Susceptibility To, 5 (CACNA1S)	1q32	CACNA1S	Seq. hot-spot gDNA
Menkes Syndrome (ATP7A)	Xq12-q13	ATP7A	Seq. gDNA
Mental Retardation, X-Linked, ARX-Related (ARX)	Xp22.13	ARX	Seq. hot-spot gDNA
Metachromatic Leukodystrophy (ARSA)	22q13.31-qter	ARSA	Seq. gDNA
Metachromatic Leukodystrophy (PSAP)	10q22.1	PSAP	Seq. gDNA
Mevalonic Aciduria (MVK)	12q24	MVK	Seq. gDNA
Mitochondrial Myopathy, Encephalopathy, Lactic Acidosis, And Stroke-Like Episod	mtDNA	-	Seq. hot-spot mtDNA
Multiple Sulfatase Deficiency (SUMF1)	3p26	SUMF1	Seq. gDNA

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Myoclonic Epilepsy Associated With Ragged-Red Fibers (-)	mtDNA	-	Seq. hot-spot mtDNA
Myotonia, Potassium-Aggravated (SCN4A)	17q23.1-q25.3	SCN4A	Seq. gDNA
Neuropathy, Ataxia, And Retinitis Pigmentosa (MTATP6)	mtDNA	MTATP6	Seq. hot-spot mtDNA
Niemann-Pick Disease, Type A (SMPD1)	11p15.4-p15.1	SMPD1	Seq. gDNA
Niemann-Pick Disease, Type B (SMPD1)	11p15.4-p15.1	SMPD1	Seq. gDNA
Niemann-Pick Disease, Type C1 (NPC1)	18q11-q12	NPC1	Seq. gDNA
Ornithine Aminotransferase Deficiency (OAT)	10q26	OAT	Seq. gDNA
Paramyotonia Congenita Of Von Eulenburg (SCN4A)	17q23.1-q25.3	SCN4A	Seq. gDNA
Paraplegia Spastica Tipo 2 (PLP1)	Xq22	PLP1	Seq. gDNA
Parkinson - Dementia Syndrome (MAPT)	17q21.1	MAPT	Seq. gDNA
Parkinson Disease 1, Familial (SNCA)	4q21	SNCA	Seq. gDNA
Parkinson Disease 13 (HTRA2)	2p12	HTRA2	Seq. gDNA
Parkinson Disease 2, Autosomal Recessive Juvenile (PARK2)	6q25.2-q27	PARK2	MLPA
Parkinson Disease 2, Autosomal Recessive Juvenile (PARK2)	6q25.2-q27	PARK2	Seq. gDNA
Parkinson Disease 4, Autosomal Dominant Lewy Body (SNCA)	4q21	SNCA	MLPA
Parkinson Disease 5 (UCHL1)	4p14	UCHL1	Seq. gDNA
Parkinson Disease 6, Autosomal Recessive Early-Onset (PINK1)	1p36	PINK1	Seq. gDNA
Parkinson Disease 7, Autosomal Recessive Early-Onset (PARK7)	1p36	PARK7	Seq. gDNA
Parkinson Disease 8 (LRRK2)	12q12	LRRK2	Seq. hot-spot gDNA
Pelizaeus-Merzbacher Disease (PLP1)	Xq22	PLP1	qPCR
Pelizaeus-Merzbacher Disease (PLP1)	Xq22	PLP1	Seq. gDNA
Pendred Syndrome (SLC26A4)	7q22.3	SLC26A4	Seq. gDNA
Prader-Willi Syndrome (SNRPN)	15q11-q13	SNRPN	MS-MLPA
Prader-Willi Syndrome (SNRPN)	15q11-q13	SNRPN	Det. UPD

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Disease Name	Locus	Gene	Method
Prader-Willi Syndrome (SNRPN)	15q11-q13	SNRPN	MS-PCR
Pseudoxanthoma Elasticum (ABCC6)	16p13.1	ABCC6	MLPA
Pseudoxanthoma Elasticum (ABCC6)	16p13.1	ABCC6	Seq. gDNA
Renal Tubular Acidosis, Distal, Autosomal Dominant (SLC4A1)	17q21-q22	SLC4A1	Seq. gDNA
Renal Tubular Acidosis, Distal, With Hemolytic Anemia (SLC4A1)	17q21-q22	SLC4A1	Seq. gDNA
Rett Syndrome (FOXG1)	14q13	FOXG1	Seq. gDNA
Rett Syndrome (MECP2)	Xq28	MECP2	MLPA
Rett Syndrome (MECP2)	Xq28	MECP2	Seq. gDNA
Spastic Ataxia, Charlevoix - Saguenay Type (SACS)	13q12	SACS	Seq. gDNA
Spastic Paraplegia 17, Autosomal Dominant (BSCL2)	11q13	BSCL2	Seq. gDNA
Spastic Paraplegia 3, Autosomal Dominant (ATL1)	14q11-q21	ATL1	Seq. gDNA
Spastic Paraplegia 3, Autosomal Dominant (ATL1)	14q11-q21	ATL1	MLPA
Spastic Paraplegia 4, Autosomal Dominant (SPAST)	2p22-p21	SPAST	MLPA
Spastic Paraplegia 4, Autosomal Dominant (SPAST)	2p22-p21	SPAST	Seq. gDNA
Spastic Paraplegia 7, Autosomal Recessive (SPG7)	16q24.3	SPG7	Seq. gDNA
Spherocytosis, Type 4 (SLC4A1)	17q21-q22	SLC4A1	Seq. gDNA
Spinal And Bulbar Muscular Atrophy, X-Linked 1 (AR)	Xq11-q12	AR	STR
Spinal And Bulbar Muscular Atrophy, X-Linked 1 (AR)	Xq11-q12	AR	RP-PCR
Spinal Muscular Atrophy, Type I (SMN1)	5q12.2-q13.3	SMN1	Seq. gDNA
Spinal Muscular Atrophy, Type I (SMN1)	5q12.2-q13.3	SMN1	MLPA
Spinocerebellar Ataxia Type 1 (ATXN1)	6p23	ATXN1	STR
Spinocerebellar Ataxia Type 10 (ATXN10)	22q13	ATXN10	STR
Spinocerebellar Ataxia Type 10 (ATXN10)	22q13	ATXN10	RP-PCR
Spinocerebellar Ataxia Type 12 (PPP2R2B)	5q31-q33	PPP2R2B	STR

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Spinocerebellar Ataxia Type 14 (PRKCG)	19q13.4	PRKCG	Seq. gDNA
Spinocerebellar Ataxia Type 17 (TBP)	6q27	TBP	STR
Spinocerebellar Ataxia Type 2 (ATXN2)	12q24	ATXN2	RP-PCR
Spinocerebellar Ataxia Type 2 (ATXN2)	12q24	ATXN2	STR
Spinocerebellar Ataxia Type 27 (FGF14)	13q34	FGF14	Seq. gDNA
Spinocerebellar Ataxia Type 3 (ATXN3)	14q24.3-q31	ATXN3	STR
Spinocerebellar Ataxia Type 36 (NOP56)	20p13	NOP56	STR
Spinocerebellar Ataxia Type 36 (NOP56)	20p13	NOP56	RP-PCR
Spinocerebellar Ataxia Type 5 (SPTBN2)	11q13	SPTBN2	Seq. gDNA
Spinocerebellar Ataxia Type 6 (CACNA1A)	19p13	CACNA1A	STR
Spinocerebellar Ataxia Type 7 (ATXN7)	3p21.1-p12	ATXN7	RP-PCR
Spinocerebellar Ataxia Type 7 (ATXN7)	3p21.1-p12	ATXN7	STR
Spinocerebellar Ataxia Type 8 (ATXN8OS)	13q21	ATXN8OS	RP-PCR
Spinocerebellar Ataxia Type 8 (ATXN8OS)	13q21	ATXN8OS	STR
Spinocerebellar Ataxia, Autosomal Recessive 1 (SETX)	9q34	SETX	Seq. gDNA
Thrombophilia (F2)	11p11-q12	F2	Seq. hot-spot gDNA
Thrombophilia Due To Activated Protein C Resistance (F5)	1q23	F5	Seq. hot-spot gDNA
TP53 Associated Diseases (TP53)	17p13.1	TP53	Seq. gDNA
Tuberous Sclerosis (TSC1)	9q34	TSC1	Seq. gDNA
Tuberous Sclerosis (TSC2)	16p13,3	TSC2	Seq. gDNA
Tuberous Sclerosis (TSC2)	16p13,3	TSC2	MLPA
Tuberous Sclerosis (TSC2)	16p13,3	TSC2	Seq. cDNA
Waardenburg Syndrome, Type I (PAX3)	2q35	PAX3	Seq. gDNA
Wilson Disease (ATP7B)	13q14.3-q21.1	ATP7B	Seq. gDNA

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Disease Name	Locus	Gene	Method
Wolfram Syndrome Type 2 (CISD2)	4q22-q24	CISD2	Seq. gDNA
Yellow Nail Syndrome (FOXC2)	16q24.3	FOXC2	Seq. gDNA