Test List

Panels Offered		
Test Code	**Panel - Test Description**	**CPT Codes**
	3-Methyl-Crotonyl-CoA Carboxylase Deficiency - 3MCC A+B	
L7464	2 Gene Sanger Sequencing (MCCC1 & MCCC2)	81406x2
MG6052	2 Gene Del-Dup Exonic Microarray (MCCC1 & MCCC2)	81479x2
	46, XY Disorder of Sexual Development	
L7332	3 Gene Sanger Sequencing (AR, SRY, WT1)	81400 & 81405x2
	Aicardi-Goutieres Syndrome (AGS)	
MG93	5 Gene Sanger Sequencing (TREX1, RNASEH2B, RNASEH2C, RNASEH2A, SAMHD1)	81479x5
L6921	5 Gene Del-Dup Exonic Microarray (TREX1, RNASEH2B, RNASEH2C, RNASEH2A, SAMHD1)	81479x5
	Albinism/Oculocutaneous Albinism: OA1, OCA1, OCA2 and OCA2 deletion, OCA3, OCA4	
L6723	5 Gene Sanger Sequencing (OA1, OCA1, OCA2, OCA3, OCA4)	81404 & 81479x4
L6724	5 Gene Del-Dup Exonic Microarray (OA1, OCA1, OCA2, OCA3, OCA4)	81479x5
	Arrhythmogenic Right Ventricular Dysplasia	
MG16	4 Gene Sanger Sequencing (PKP2, DSP, DSG2, DSC2)	81406x4
MG6016	4 Gene Del-Dup Exonic Microarray (PKP2, DSP, DSG2, DSC2)	81406x4
MG16C	1 Known Mutation in the 4 ARVD	81479
	Chronic Granulomatous Disease	
MG284	3 Gene Sanger Sequencing (CYBA, CYBB, NCF2)	81479x3
MG6284	3 Gene Del-dup Exonic Microarray (CYBA, CYBB, NCF2)	81479x3
	Fibroblast Growth Factor Receptor Disorders - FGFR	
L7039	3 Gene Sanger Sequencing (FGFR1, FGFR2, FGFR3)	81479x3
L6926	3 Gene Del-Dup Exonic Microarray (FGFR1, FGFR2, FGFR3)	81479x3
MG241P	3 Gene Prenatal Sanger Sequencing incl. MCC (FGFR1, FGFR2, FGFR3)	81479x3
	Glutaric Acidemia Type 2 - GA2	
L6936	3 Gene Sanger Sequencing (ETFA, ETFB, ETFDH)	81479x3
MG6057	3 Gene Del-Dup Exonic Microarray (ETFA, ETFB, ETFDH)	81479x3
	Methylmalonic Acidemia - MMA	
L7052	24 Gene NextGen Sequencing Panel	81401, 81406x3, 81405x2, 81479x18
MG66	3 Gene Sanger Sequencing (MMAA, MMAB, MUT)	81479x3
MG6066	3 Gene Del-Dup Exonic Microarray (MMAA, MMAB, MUT)	81479x3
	Non-Ketotic Hyperglycinemia (Glycine Encephalopathy) - NKH	
L6928	3 Gene Sanger Sequencing (AMT, GCSH, GLDC)	81479x3
L7152	3 Gene Del-Dup Exonic Microarray (AMT, GCSH, GLDC)	81479x3
	Propionic Acidemia - PCC	
MG77	2 Gene Sanger Sequencing (PCCA, PCCB)	81406x2
MG6077	2 Gene Del-Dup Exonic Microarray (PCCA, PCCB)	81405 & 81479
	RASopathy Spectrum Disorders (Noonan)	
L7273	NextGen Sequencing (NGS)	81442
MG6292	Single Gene Del-Dup OGT Microarray	81323
L7424	1 or 2 Known Mutation	81322
MG292P	Prenatal NGS incl. MCC	81442
MG292P1	Prenatal 1 Known Mutation incl. MCC	81322
MG292P2	Prenatal 2 Known Mutation incl. MCC	81323

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	Rubinstein-Taybi Syndrome	
L7124	2 Gene Sanger Sequencing (CREBBP, EP300)	81407 & 81479
L7125	2 Gene Del-Dup Exonic Microarray (CREBBP, EP300)	81406 & 81479
	Waardenburg Syndrome	
MG32	4 Gene Sanger Sequencing (EDNRB, MITF, PAX3, SOX10)	81479x4

Individual Genes Offered

Test Code	Gene - Test Description	CPT
	ABCA4 - Stargardt, arRP, Age-Related Macular Degenration, CORD3	
MG124	Sanger Sequencing	81408
MG6124	Del-Dup Exonic Microarray	81479
MG124C	1 or 2 Known Mutation	81479
MG124P	Prenatal Sequencing incl. MCC	81408
MG124P1	Prenatal 1 Known Mutation incl. MCC	81479
MG124P2	Prenatal 2 Known Mutation incl. MCC	81479
	ACADM - Medium Chain Acyl-CoA Dehydrogenase Deficiency, MCAD	
L6917	Sanger Sequencing	81479
L7279	Sanger Sequencing Common Mutation (K304E)	81400
L7277	Del-Dup Exonic Microarray	81479
L7278	1 or 2 Known Mutation	81479
MG65P	Prenatal Sequencing incl. MCC	81479
MG65P1	Prenatal 1 Known Mutation incl. MCC	81479
MG65P2	Prenatal 2 Known Mutation incl. MCC	81479
	ACADVL - Very Long Chain Acyl-CoA Dehydrogenase Deficiency, VLCAD	
L6906	Sanger Sequencing	81406
L7161	Del-Dup Exonic Microarray	81479
L7206	1 or 2 Known Mutation	81403
MGP	Prenatal Sequencing incl. MCC	81406
MGP1	Prenatal 1 Known Mutation incl. MCC	81479
MGP2	Prenatal 2 Known Mutation incl. MCC	81479
	ACVRL1/ALK1 - Hereditary Hemorrhagic Telangiectasia	
MG262	Sanger Sequencing	81479
MG6262	Del-Dup Exonic Microarray	81479
MG262C	1 or 2 Known Mutation	81479
MG262P	Prenatal Sequencing incl. MCC	81479
MG262P1	Prenatal 1 Known Mutation incl. MCC	81479
MG262P2	Prenatal 2 Known Mutation incl. MCC	81479
	ALDH7A1 - Antiquitin, Pyridoxine-Dependent Neonatal Seizures, ATQ-1	
MG55	Sanger Sequencing	81406
MG6055	Del-Dup Exonic Microarray	81479
MG55C	1 or 2 Known Mutation	81479
MG55P	Prenatal Sequencing incl. MCC	81406
MG55P1	Prenatal 1 Known Mutation incl. MCC	81479
MG55P2	Prenatal 2 Known Mutation incl. MCC	81479

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	ALX4 - Frontonasal Dysplasia 2; Parietal Foramina 2	
MG347	Sanger Sequencing	81479
MG6347	Del-Dup Exonic Microarray	81479
MG347C	1 or 2 Known Mutation	81479
MG347P	Prenatal Sequencing incl. MCC	81479
MG347P1	Prenatal 1 Known Mutation incl. MCC	81479
MG347P2	Prenatal 2 Known Mutation incl. MCC	81479
	AMT - Non-Ketotic Hyperglycinemia, Glycine Encephalopathy, NKH	
L7153	Sanger Sequencing	81479
L7154	Del-Dup Exonic Microarray	81479
L7207	1 or 2 Known Mutation	81479
MG74P	Prenatal Sequencing incl. MCC	81479
MG74P1	Prenatal 1 Known Mutation incl. MCC	81479
MG74P2	Prenatal 2 Known Mutation incl. MCC	81479
	APC - Familial adenomatous polyposis (FAP)/Gardner syndrome/Turcot Syndrome	
MG267	Sanger Sequencing	81201
MG6267	Del-Dup Exonic Microarray	81203
MG267C	1 or 2 Known Mutation	81202
MG267P	Prenatal Sequencing incl. MCC	81201
MG267P1	Prenatal 1 Known Mutation incl. MCC	81202
MG267P2	Prenatal 2 Known Mutation incl. MCC	81202
	AR - 46, XY Disorder of Sexual Development	
L7174	Sanger Sequencing	81405
L7290	Del-Dup Exonic Microarray	81479
L7291	1 or 2 Known Mutation	81479
MG38P	Prenatal Sequencing incl. MCC	81405
MG38P1	Prenatal 1 Known Mutation incl. MCC	81479
MG38P2	Prenatal 2 Known Mutation incl. MCC	81479
	BAG3 - Dilated Cardiomyopathy; Myofibrillar Myopathy; Primary Dilated Cardiomyopathy	
MG348	Sanger Sequencing	81479
MG348C	1 or 2 Known Mutation	81479
MG348P	Prenatal Sequencing incl. MCC	81479
MG348P1	Prenatal 1 Known Mutation incl. MCC	81479
MG348P2	Prenatal 2 Known Mutation incl. MCC	81479
	BTD - Biotinidase Deficiency	
L7268	Sanger Sequencing	81404
L7359	Del-Dup Exonic Microarray	81479
MG126C	1 or 2 Known Mutation	81479
MG126P	Prenatal Sequencing incl. MCC	81404
MG126P1	Prenatal 1 Known Mutation incl. MCC	81479
MG126P2	Prenatal 2 Known Mutation incl. MCC	81479
	BCKDHA Y438N & BCKDHB R183P, G278S & E372X - Maple Syrup Disease - Common Jewish Mutations	
MG100	Sanger Sequencing	81479
MG100C	1 or 2 Known Mutation	81479
	BOLA3 - Multiple Mitochondrial Dysfunctions Syndrome, MMDS	
MG243	Sanger Sequencing	81479
MG6243	Del-Dup Exonic Microarray	81479
MG243C	1 or 2 Known Mutation	81479
MG243P	Prenatal Sequencing incl. MCC	81479
MG243P1	Prenatal 1 Known Mutation incl. MCC	81479

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MG243P2	Prenatal 2 Known Mutation incl. MCC	81479

	C-KIT - Piebaldism, Mast Cell Leukemia, GIST	
MG105	Sanger Sequencing	81479
MG6105	Del-Dup Exonic Microarray	81479
MG105C	1 or 2 Known Mutation	81479
MG105P	Prenatal Sequencing incl. MCC	81479
MG105P1	Prenatal 1 Known Mutation incl. MCC	81479
MG105P2	Prenatal 2 Known Mutation incl. MCC	81479
	C7orf10 - Glutaric Acidemia Type 3, GA3	
L7300	Sanger Sequencing	81479
L7483	Del-Dup Exonic Microarray	81479
MG102C	1 or 2 Known Mutation	81479
MG102P	Prenatal Sequencing incl. MCC	81479
MG102P1	Prenatal 1 Known Mutation incl. MCC	81479
MG102P2	Prenatal 2 Known Mutation incl. MCC	81479
	CBS - Homocystinuria due to Cystathionine Beta-Synthase Deficiency	
L7346	Sanger Sequencing	81406
L7286	Del-Dup Exonic Microarray	81479
L7287	1 or 2 Known Mutation	81479
MG62P	Prenatal Sequencing incl. MCC	81406
MG62P1	Prenatal 1 Known Mutation incl. MCC	81479
MG62P2	Prenatal 2 Known Mutation incl. MCC	81479
	CDKL5 - Atypical Rett Syndrome	
MG260	Sanger Sequencing	81406
MG6260	Del-Dup Exonic Microarray	81405
MG260C	1 or 2 Known Mutation	81479
MG260P	Prenatal Sequencing incl. MCC	81406
MG260P1	Prenatal 1 Known Mutation incl. MCC	81479
MG261P2	Prenatal 2 Known Mutation incl. MCC	81479
	CEP290 - Joubert Syndrome, Bardet-Biedl, Meckel, Senior-Loken, LCA	
MG107	Sanger Sequencing	81408
MG6107	Del-Dup Exonic Microarray	81479
MG107C	1 or 2 Known Mutation	81479
MG107P	Prenatal Sequencing incl. MCC	81408
MG107P1	Prenatal 1 Known Mutation incl. MCC	81479
MG107P2	Prenatal 2 Known Mutation incl. MCC	81479
	CFTR - Cystic Fibrosis	
L7050	NextGen Sequencing	81223
L6935	23 Common Variants NextGen Sequencing	81220
L7051	Del-Dup Exonic Microarray	81222
L7242	1 or 2 Known Mutation	81221
MG14P	Prenatal Gene Analysis; 23 Common Variants incl. MCC	81220
	CHD7 - Charge Syndrome	
L6913	Sanger Sequencing	81407
L7324	Del-Dup Exonic Microarray	81479
MG272C	1 or 2 Known Mutation	81479
MG272P	Prenatal Sequencing incl. MCC	81407
MG272P1	Prenatal 1 Known Mutation incl. MCC	81479
MG272P2	Prenatal 2 Known Mutation incl. MCC	81479

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	CHS1 (LYST) - Chediak-Higashi Syndrome	
MG83	Sanger Sequencing	81479
MG6083	Del-Dup Exonic Microarray	81479
MG83C	1 or 2 Known Mutation	81479
MG83P	Prenatal Sequencing incl. MCC	81479
MG83P1	Prenatal 1 Known Mutation incl. MCC	81479
MG83P2	Prenatal 2 Known Mutation incl. MCC	81479
	CNGA3 - Achromatopsia	
MG119	Sanger Sequencing	81479
MG6119	Del-Dup Exonic Microarray	81479
MG119C	1 or 2 Known Mutation	81479
MG119P	Prenatal Sequencing incl. MCC	81479
MG119P1	Prenatal 1 Known Mutation incl. MCC	81479
MG119P2	Prenatal 2 Known Mutation incl. MCC	81479
	CNGB3 - Achromatopsia	
MG120	Sanger Sequencing	81479
MG6120	Del-Dup Exonic Microarray	81479
MG120C	1 or 2 Known Mutation	81479
MG120P	Prenatal Sequencing incl. MCC	81479
MG120P1	Prenatal 1 Known Mutation incl. MCC	81479
MG120P2	Prenatal 2 Known Mutation incl. MCC	81479
	CREBBP - Rubinstein-Taybi Syndrome	
L6908	Sanger Sequencing	81407
L7271	Del-Dup Exonic Microarray	81406
L7427	1 or 2 Known Mutation	81479
MG112P	Prenatal Sequencing incl. MCC	81407
MG112P1	Prenatal 1 Known Mutation incl. MCC	81479
MG112P2	Prenatal 2 Known Mutation incl. MCC	81479
	CYBA - Chronic Granulomatous Disease	
MG152	Sanger Sequencing, p22-PHOX	81479
MG6152	Del-Dup Exonic Microarray	81479
MG152C	1 or 2 Known Mutation	81479
MG152P	Prenatal Sequencing incl. MCC	81479
MG152P1	Prenatal 1 Known Mutation incl. MCC	81479
MG152P2	Prenatal 2 Known Mutation incl. MCC	81479
	CYBB - Chronic Granulomatous Disease	
L6922	Sanger Sequencing, p91-PHOX	81479
L7340	Del-Dup Exonic Microarray	81479
L7428	1 or 2 Known Mutation	81479
MG151P	Prenatal Sequencing incl. MCC	81479
MG151P1	Prenatal 1 Known Mutation incl. MCC	81479
MG151P2	Prenatal 2 Known Mutation incl. MCC	81479
	DSC2 - Arrhythmogenic Right Ventricular Dysplasia (ARVD11)	
MG20	Sanger Sequencing	81406
MG6020	Del-Dup Exonic Microarray	81479
MG20C	1 or 2 Known Mutation	81479
MG20P	Prenatal Sequencing incl. MCC	81406
MG20P1	Prenatal 1 Known Mutation incl. MCC	81479

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MG20P2	Prenatal 2 Known Mutation incl. MCC	81479

	DSG2 - Arrhythmogenic Right Ventricular Dysplasia (ARVD10)	
MG20	Sanger Sequencing	81406
MG6020	Del-Dup Exonic Microarray	81479
MG20C	1 or 2 Known Mutation	81479
MG20P	Prenatal Sequencing incl. MCC	81406
MG20P1	Prenatal 1 Known Mutation incl. MCC	81479
MG20P2	Prenatal 2 Known Mutation incl. MCC	81479
	DSP- Arrhthmogenic Right Ventricular Dysplasia (ARVD8)	
MG19	Sanger Sequencing	81406
MG6019	Del-Dup Exonic Microarray	81479
MG19C	1 or 2 Known Mutation	81479
MG19P	Prenatal Sequencing incl. MCC	81406
MG19P1	Prenatal 1 Known Mutation incl. MCC	81479
MG19P2	Prenatal 2 Known Mutation incl. MCC	81479
	DHCR7 - Smith Lemli Opitz, SLOS	
L6909	Sanger Sequencing	81405
L7288	Del-Dup Exonic Microarray	81479
L7289	1 or 2 Known Mutation	81479
MG136P	Prenatal Sequencing incl. MCC	81405
MG136P1	Prenatal 1 Known Mutation incl. MCC	81479
MG136P2	Prenatal 2 Known Mutation incl. MCC	81479
	DHH - 46, XY Disorder of Sexual Development	
MG349	Sanger Sequencing	81479
MG6349	Del-Dup Exonic Microarray	81479
MG349C	1 or 2 Known Mutation	81479
MG349P	Prenatal Sequencing incl. MCC	81479
MG349P1	Prenatal 1 Known Mutation incl. MCC	81479
MG349P2	Prenatal 2 Known Mutation incl. MCC	81479
	EDN3 - Waardenburg Syndrome	
MG254	Sanger Sequencing	81479
MG6254	Del-Dup Exonic Microarray	81479
MG254C	1 or 2 Known Mutation	81479
MG254P	Prenatal Sequencing incl. MCC	81479
MG254P1	Prenatal 1 Known Mutation incl. MCC	81479
MG254P2	Prenatal 2 Known Mutation incl. MCC	81479
	EDNRB - Waardenburg Syndrome	
MG36	Sanger Sequencing	81479
MG6036	Del-Dup Exonic Microarray	81479
MG36C	1 or 2 Known Mutation	81479
MG36P	Prenatal Sequencing incl. MCC	81479
MG36P1	Prenatal 1 Known Mutation incl. MCC	81479
MG36P2	Prenatal 2 Known Mutation incl. MCC	81479
	EFNB1 - Craniofrontonasal Dysplasia	
L6923	Sanger Sequencing	81479
MG6253	Del-Dup Exonic Microarray	81479
MG253C	1 or 2 Known Mutation	81479

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MG253P	Prenatal Sequencing incl. MCC	81479
MG253P1	Prenatal 1 Known Mutation incl. MCC	81479
MG253P2	Prenatal 2 Known Mutation incl. MCC	81479

	EFTUD2 - Mandibulofacial Dysostosis with Microcephaly	
L7325	Sanger Sequencing	81479
L7384	Del-Dup Exonic Microarray	81479
L7386	1 or 2 Known Mutation	81479
MG304P	Prenatal Sequencing incl. MCC	81479
MG304P1	Prenatal 1 Known Mutation incl. MCC	81479
MG304P2	Prenatal 2 Known Mutation incl. MCC	81479
	ELANE - Severe Congenital Neutropenia and Cyclic Neutropenia	
MG306	Sanger Sequencing	81479
MG6306	Del-Dup Exonic Microarray	81479
MG306C	1 or 2 Known Mutation	81479
MG306P	Prenatal Sequencing incl. MCC	81479
MG306P1	Prenatal 1 Known Mutation incl. MCC	81479
MG306P2	Prenatal 2 Known Mutation incl. MCC	81479
	ENG - Hereditary Hemorrhagic Telangiectasia	
MG263	Sanger Sequencing	81406
MG6263	Del-Dup Exonic Microarray	81405
MG263C	1 or 2 Known Mutation	81479
MG263P	Prenatal Sequencing incl. MCC	81406
MG263P1	Prenatal 1 Known Mutation incl. MCC	81479
MG263P2	Prenatal 2 Known Mutation incl. MCC	81479
	EP300 - Rubenstein-Taybi Syndrome	
L7267	Sanger Sequencing	81479
L7266	Del-Dup Exonic Microarray	81479
L7341	1 or 2 Known Mutation	81479
MG113P	Prenatal Sequencing incl. MCC	81479
MG113P1	Prenatal 1 Known Mutation incl. MCC	81479
MG113P2	Prenatal 2 Known Mutation incl. MCC	81479
	ETFA - Glutaric Acidemia Type 2, GA2	
L7305	Sanger Sequencing	81479
L7306	Del-Dup Exonic Microarray	81479
L7307	1 or 2 Known Mutation	81479
MG59P	Prenatal Sequencing incl. MCC	81479
MG59P1	Prenatal 1 Known Mutation incl. MCC	81479
MG59P2	Prenatal 2 Known Mutation incl. MCC	81479
	ETFB - Glutaric Acidemia Type 2, GA2	
L7308	Sanger Sequencing	81479
L7309	Del-Dup Exonic Microarray	81479
L7310	1 or 2 Known Mutation	81479
MG60P	Prenatal Sequencing incl. MCC	81479
MG60P1	Prenatal 1 Known Mutation incl. MCC	81479
MG60P2	Prenatal 2 Known Mutation incl. MCC	81479
	ETFDH - Glutaric Acidemia Type 2, GA2	
L7311	Sanger Sequencing	81479

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L7312	Del-Dup Exonic Microarray	81479
L7313	1 or 2 Known Mutation	81479
MG58P	Prenatal Sequencing incl. MCC	81479
MG58P1	Prenatal 1 Known Mutation incl. MCC	81479
MG58P2	Prenatal 2 Known Mutation incl. MCC	81479

	FGFR1 - Pfeiffer Syndrome, Osteoglophonic Dysplasia	
L7262	Sanger Sequencing	81405
L7347	Sanger Sequencing for P252R Mutation - Pfeiffer	81400
L7345	Del-Dup Exonic Microarray	81479
MG128C	1 or 2 Known Mutation	81479
MG128P	Prenatal Sequencing incl. MCC	81405
MG128P1	Prenatal 1 Known Mutation incl. MCC	81479
MG128P2	Prenatal 2 Known Mutation incl. MCC	81479
	FGFR2 - Crouzon, Jackson-Weiss, Beare-Stevenson, Apert, Pfeiffer	
L7046	Sanger Sequencing	81404
L7282	Del-Dup Exonic Microarray	81479
L7283	1 or 2 Known Mutation	81479
MG129P	Prenatal Sequencing incl. MCC	81404
MG129P1	Prenatal 1 Known Mutation incl. MCC	81479
MG129P2	Prenatal 2 Known Mutation incl. MCC	81479
	FGFR2 - Apert Specific	
L7048	Sanger Sequencing for S252W, P253R Mutations	81404
MG236P1	Prenatal 1 Known Mutation incl. MCC	81404
MG236P2	Prenatal 2 Known Mutation incl. MCC	81404
	FGFR2 - Beare-Stevenson Specific	
L7344	Sanger Sequencing for S372C, Y375C Mutations	81404
MG237P1	Prenatal 1 Known Mutation incl. MCC	81404
MG237P2	Prenatal 2 Known Mutation incl. MCC	81479
	FGFR3 - Achondroplasia, Hypochondroplasia, Thanaophoric Dysplasia, SADDAN, Craniosyostoses, Crouzon, Muenke	
L6907	Sanger Sequencing	81479
L7272	Del-Dup Exonic Microarray	81479
L7263	1 or 2 Known Mutation	81403
MG130P	Prenatal Sequencing incl. MCC	81479
MG130P1	Prenatal 1 Known Mutation incl. MCC	81479
MG130P2	Prenatal 2 Known Mutation incl. MCC	81479
L7299	Tier 1 (sanger sequencing exons 7, 9, 12, 14 & 18)	81403, 81404
LF1054	Prenatal Tier 1 incl. MCC	81404, 81479
L7264	Add-On Sequencing *after* Tier 1	81479
MG142P	Prenatal Add-On Sequencing *after* Tier 1 incl. MCC	81479
	FGFR3 - Crouzon (A391E) Syndrome Specific	
L7338	Sanger Sequencing	81404
MG44P	Prenatal Sequencing incl. MCC	81404
	FGFR3 - Muenke (P250R Variant) Syndrome Specific	
L6914	Sanger Sequencing	81400
MG43P	Prenatal Sequencing incl. MCC	81400
	FGFR3 - Saddan (K650M) Specific	
L7304	Sanger Sequencing	81404

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MG45P	Prenatal Sequencing incl. MCC	81404
	FMO3 - Trimethylaminuria, TMA	
L6927	Sanger Sequencing	81479
L7284	Del-Dup Exonic Microarray	81479
L7285	1 or 2 Known Mutation	81479
	FMR1 - Fragile X: FX	
L6943	CGG repeats, FX (includes Southern Blot if positive)	81243 & 81244
L7204	Del-Dup Exonic Microarray	81479
MG46P	Prenatal CGG Repeats, FX incl. MCC (includes Southern Blot if positive)	81243, 81244

	GCDH - Glutaric Acidemia Type 1, GA1	
L6905	Sanger Sequencing	81406
L7160	Del-Dup Exonic Microarray	81479
L7205	1 or 2 Known Mutation	81403
MG56P	Prenatal Sequencing incl. MCC	81406
MG56P1	Prenatal 1 Known Mutation incl. MCC	81403
MG56P2	Prenatal 2 Known Mutation incl. MCC	81403
	GCSH - Non-Ketotic Hyperglycinemia, Glycine Encephalopathy, NKH	
L7157	Sanger Sequencing	81479
L7158	Del-Dup Exonic Microarray	81479
L7208	1 or 2 Known Mutation	81479
MG76P	Prenatal Sequencing incl. MCC	81479
MG76P1	Prenatal 1 Known Mutation incl. MCC	81479
MG76P2	Prenatal 2 Known Mutation incl. MCC	81479
	GJB2 - Connexin 26 - Related Hearing Loss	
L6919	Sanger Sequencing	81252
MG6029	Del-Dup Exonic Microarray	81479
MG29C	1 or 2 Known Mutation	81253
MG29P	Prenatal Sequencing incl. MCC	81252
MG29P1	Prenatal 1 Known Mutation incl. MCC	81253
MG29P2	Prenatal 2 Known Mutation incl. MCC	81253
	GJB6 - Connexin 30 - Related Hearing Loss	
MG30	Common Deletion	81254
MG6030	Del-Dup Exonic Microarray	81479
MG30P1	Common Deletion Prenatal 1 Known Mutation incl. MCC	81254
	GLDC - Non-Ketotic Hyperglycinemia, Glycine Encephalopathy, NKH	
L7155	Sanger Sequencing	81479
L7156	Del-Dup Exonic Microarray	81479
L6726	1 or 2 Known Mutation	81479
L7328	Prenatal Sequencing incl. MCC	81479
L7327	Prenatal 1 Known Mutation incl. MCC	81479
L7326	Prenatal 2 Known Mutation incl. MCC	81479
	GNAI3 - Auriculocondylar Syndrome Type 1	
L7171	Sanger Sequencing	81479
L7170	Del-Dup Exonic Microarray	81479
MG305C	1 or 2 Known Mutation	81479
MG305P	Prenatal Sequencing incl. MCC	81479
MG305P1	Prenatal 1 Known Mutation incl. MCC	81479
MG305P2	Prenatal 2 Known Mutation incl. MCC	81479
	GNAT2 - Achromatopsia	
MG121	Sanger Sequencing	81479

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MG6121	Del-Dup Exonic Microarray	81479
MG121C	1 or 2 Known Mutation	81479
MG121P	Prenatal Sequencing incl. MCC	81479
MG121P1	Prenatal 1 Known Mutation incl. MCC	81479
MG121P2	Prenatal 2 Known Mutation incl. MCC	81479

	GPR143 - Albinism/Oculocutaneous Albinism: OA1 (X-linked)	
MG92	Sanger Sequencing	81479
MG6092	Del-Dup Exonic Microarray	81479
MG92C	1 or 2 Known Mutation	81479
MG92P	Prenatal Sequencing incl. MCC	81479
MG92P1	Prenatal 1 Known Mutation incl. MCC	81479
MG92P2	Prenatal 2 Known Mutation incl. MCC	81479
	HADHA - Long Chain 3-Hydroxy-Acyl-CoA Dehydrogenase Deficiency, LCHAD	
L6933	Sanger Sequencing of Common Mutation (E5100Q)	81479
L7276	Del-Dup Exonic Microarray	81479
MG63P	Prenatal Sequencing of E5100Q incl. MCC	81479
MG63P1	Prenatal 1 Known Mutation of E5100Q incl. MCC	81479
	HFE - Hereditary Hemochromatosis	
L6930	Sanger Sequencing of exons 2 & 4 to detect C282Y, H63D mutations	81256
	HLCS - Holocarboxylase Synthetase Deficiency	
MG61	Sanger Sequencing	81406
MG6061	Del-Dup Exonic Microarray	81479
MG61C	1 or 2 Known Mutation	81479
MG61P	Prenatal Sequencing incl. MCC	81406
MG61P1	Prenatal 1 Known Mutation incl. MCC	81479
MG61P2	Prenatal 2 Known Mutation incl. MCC	81479
	HPS1 - Hermansky - Pudlak Syndrome: Common Puerto Rican Mutation 1470_1486dup16	
MG84	Sanger Sequencing	81779
MG84P	Prenatal Sequencing incl. MCC	81479
	HPS3 - Hermansky - Pudlak Syndrome: Common Puerto Rican Mutation 339_4260del3904	
MG85	Sanger Sequencing	81479
MG85P	Prenatal Sequencing incl. MCC	81479
	HPS3 - Hermansky - Pudlak Syndrome: Common Ashkenazi Jewish Mutation 1163+1G>A	
MG86	Sanger Sequencing	81479
MG86P	Prenatal Sequencing incl. MCC	81479
	L1CAM - Hydrocephalus Due to Congenital Stenosis of Aqueduct of Sylvius/MASA Syndrome	
L7469	Sanger Sequencing	81407
MG6282	Del-Dup Exonic Microarray	81479
MG282C	1 or 2 Known Mutation	81479
MG282P	Prenatal Sequencing incl. MCC	81407
MG282P1	Prenatal 1 Known Mutation incl. MCC	81479
MG282P2	Prenatal 2 Known Mutation incl. MCC	81479
	LAMP2 - Danon Disease, GSD IIB (X-linked)	
MG22	Sanger Sequencing	81405

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MG6022	Del-Dup Exonic Microarray	81479
MG22C	1 or 2 Known Mutation	81479
MG22P	Prenatal Sequencing incl. MCC	81405
MG22P1	Prenatal 1 Known Mutation incl. MCC	81479
MG22P2	Prenatal 2 Known Mutation incl. MCC	81479
	LIAS - Pyruvate Dehydrogenase Lipoic Acid Synthetase Deficiency	
MG283	Sanger Sequencing	81479
MG283C	1 or 2 Known Mutation	81479
MG283P	Prenatal Sequencing incl. MCC	81479
MG283P1	Prenatal 1 Known Mutation incl. MCC	81479
MG283P2	Prenatal 2 Known Mutation incl. MCC	81479

	LMBRD1 - Cobalamin F Deficiency	
MG242	Sanger Sequencing	81479
MG6242	Del-Dup Exonic Microarray	81479
MG242C	1 or 2 Known Mutation	81479
MG242P	Prenatal Sequencing incl. MCC	81479
MG242P1	Prenatal 1 Known Mutation incl. MCC	81479
MG242P2	Prenatal 2 Known Mutation incl. MCC	81479
	LMNA - Lamin A/C	
MG23	Sanger Sequencing	81406
MG6023	Del-Dup Exonic Microarray	81479
L7339	1 or 2 Known Mutation	81479
MG23P	Prenatal Sequencing incl. MCC	81406
MG23P1	Prenatal 1 Known Mutation incl. MCC	81479
MG23P2	Prenatal 2 Known Mutation incl. MCC	81479
	MCCC1 - 3-Methyl-Crotonyl-CoA Carboxylase Deficiency	
MG53	Sanger Sequencing	81406
MG6053	Del-Dup Exonic Microarray	81479
MG53C	1 or 2 Known Mutation	81479
MG53P	Prenatal Sequencing incl. MCC	81406
MG53P1	Prenatal 1 Known Mutation incl. MCC	81479
MG53P2	Prenatal 2 Known Mutation incl. MCC	81479
	MCCC2 - 3-Methyl-Crotonyl-CoA Carboxylase Deficiency	
MG54	Sanger Sequencing	81406
MG6054	Del-Dup Exonic Microarray	81479
MG54C	1 or 2 Known Mutation	81479
MG54P	Prenatal Sequencing incl. MCC	81406
MG54P1	Prenatal 1 Known Mutation incl. MCC	81479
MG54P2	Prenatal 2 Known Mutation incl. MCC	81479
	MCEE - Epimerase Deficiency	
MG72	Sanger Sequencing	81479
MG6072	Del-Dup Exonic Microarray	81479
MG72C	1 or 2 Known Mutation	81479
MG72P	Prenatal Sequencing incl. MCC	81479
MG72P1	Prenatal 1 Known Mutation incl. MCC	81479
MG72P2	Prenatal 2 Known Mutation incl. MCC	81479
	MECP2 - Rett Syndrome	
L7335	Sanger Sequencing	81302
L7336	Del-Dup Exonic Microarray	81304

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MG285C	1 or 2 Known Mutation	81303
MG285P	Prenatal Sequencing incl. MCC	81302
MG285P1	Prenatal 1 Known Mutation incl. MCC	81303
MG285P2	Prenatal 2 Known Mutation incl. MCC	81303
	MEN1 - Multiple Endocrine Neoplasia Type 1	
L7581	Sanger Sequencing	81405
MG6286	Del-Dup Exonic Microarray	81404
MG286C	1 or 2 Known Mutation	81479
MG286P	Prenatal Sequencing incl. MCC	81405
MG286P1	Prenatal 1 Known Mutation incl. MCC	81479
MG286P2	Prenatal 2 Known Mutation incl. MCC	81479
	MITF - Waardenburg Syndrome	
MG34	Sanger Sequencing	81479
MG6034	Del-Dup Exonic Microarray	81479
MG34C	1 or 2 Known Mutation	81479
MG34P	Prenatal Sequencing incl. MCC	81479
MG34P1	Prenatal 1 Known Mutation incl. MCC	81479
MG34P2	Prenatal 2 Known Mutation incl. MCC	81479
	MMAA - Methylmalonic Acidemia	
MG68	Sanger Sequencing	81405
MG6068	Del-Dup Exonic Microarray	81479
MG68C	1 or 2 Known Mutation	81479
MG68P	Prenatal Sequencing incl. MCC	81405
MG68P1	Prenatal 1 Known Mutation incl. MCC	81479
MG68P2	Prenatal 2 Known Mutation incl. MCC	81479
	MMAB - Methylmalonic Acidemia	
MG69	Sanger Sequencing	81405
MG6069	Del-Dup Exonic Microarray	81479
MG69C	1 or 2 Known Mutation	81479
MG69P	Prenatal Sequencing incl. MCC	81405
MG69P1	Prenatal 1 Known Mutation incl. MCC	81479
MG69P2	Prenatal 2 Known Mutation incl. MCC	81479
	MMACHC - Cobalamin C Deficiency	
MG71	Sanger Sequencing	81404
MG6071	Del-Dup Exonic Microarray	81479
MG71C	1 or 2 Known Mutation	81479
MG71P	Prenatal Sequencing incl. MCC	81404
MG71P1	Prenatal 1 Known Mutation incl. MCC	81479
MG71P2	Prenatal 2 Known Mutation incl. MCC	81479
	MMADHC (C2orf25) - Cobalamin D Deficiency	
MG251	Sanger Sequencing	81479
MG6251	Del-Dup Exonic Microarray	81479
MG251C	1 or 2 Known Mutation	81479
MG251P	Prenatal Sequencing incl. MCC	81479
MG251P1	Prenatal 1 Known Mutation incl. MCC	81479
MG251P2	Prenatal 2 Known Mutation incl. MCC	81479
	MUT - Mutase Deficiency	

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MG67	Sanger Sequencing	81406
MG6067	Del-Dup Exonic Microarray	81479
MG67C	1 or 2 Known Mutation	81479
MG67P	Prenatal Sequencing incl. MCC	81406
MG67P1	Prenatal 1 Known Mutation incl. MCC	81479
MG67P2	Prenatal 2 Known Mutation incl. MCC	81479
	NCF2 - Chronic Granulomatous Disease	
MG153	Sanger Sequencing	81479
MG6153	Del-Dup Exonic Microarray	81479
MG153C	1 or 2 Known Mutation	81479
MG153P	Prenatal Sequencing incl. MCC	81479
MG153P1	Prenatal 1 Known Mutation incl. MCC	81479
MG153P2	Prenatal 2 Known Mutation incl. MCC	81479
	NCF4 - Chronic Granulomatous Disease	
MG154	Sanger Sequencing - p40-PHOX	81479
MG6154	Del-Dup Exonic Microarray	81479
MG154C	1 or 2 Known Mutation	81479
MG154P	Prenatal Sequencing incl. MCC	81479
MG154P1	Prenatal 1 Known Mutation incl. MCC	81479
MG154P2	Prenatal 2 Known Mutation incl. MCC	81479
	NFU1 - Multiple Mitochondrial Dysfunctions Syndrome, MMDS	
MG244	Sanger Sequencing	81479
MG6244	Del-Dup Exonic Microarray	81479
MG244C	1 or 2 Known Mutation	81479
MG244P	Prenatal Sequencing incl. MCC	81479
MG244P1	Prenatal 1 Known Mutation incl. MCC	81479
MG244P2	Prenatal 2 Known Mutation incl. MCC	81479
	OTX2 - Microophthalmia, Pituitary Hormone Deficiency, Retinal Dystrophy	
MG255	Sanger Sequencing	81479
MG6255	Del-Dup Exonic Microarray	81479
MG255C	1 or 2 Known Mutation	81479
MG255P	Prenatal Sequencing incl. MCC	81479
MG255P1	Prenatal 1 Known Mutation incl. MCC	81479
MG255P2	Prenatal 2 Known Mutation incl. MCC	81479
	P-gene - Albinism/Oculocutaneous Albinism: OCA2	
MG88	Sanger Sequencing	81479
MG6088	Del-Dup Exonic Microarray	81479
MG88C	1 or 2 Known Mutation	81479
MG88P	Prenatal Sequencing incl. MCC	81479
MG88P1	Prenatal 1 Known Mutation incl. MCC	81479
MG88P2	Prenatal 2 Known Mutation incl. MCC	81479
MG147	Sanger Sequencing plus 2.7kb Deletion	81479
MG89	2.7kb Deletion Only	81479
MG89P	Prenatal 2.7kb Deletion Only, incl. MCC	81479
	PAX3 - Waardenburg Syndrome	
MG33	Sanger Sequencing	81479
MG6033	Del-Dup Exonic Microarray	81479

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MG33C	1 or 2 Known Mutation	81479
MG33P	Prenatal Sequencing incl. MCC	81479
MG33P1	Prenatal 1 Known Mutation incl. MCC	81479
MG33P2	Prenatal 2 Known Mutation incl. MCC	81479
	PAX6 - PAX6 Related Disorders: Isolated Aniridia, Anophthalmia, Gillespie	
L6948	Sanger Sequencing	81479
L6722	Del-Dup Exonic Microarray	81479
MG148C	1 or 2 Known Mutation	81479
MG148P	Prenatal Sequencing incl. MCC	81479
MG148P1	Prenatal 1 Known Mutation incl. MCC	81479
MG148P2	Prenatal 2 Known Mutation incl. MCC	81479
	PCCA - Propionic Acidemia PCC	
L7462	Sanger Sequencing	81406
MG6078	Del-Dup Exonic Microarray	81405
MG78C	1 or 2 Known Mutation	81479
MG78P	Prenatal Sequencing incl. MCC	81406
MG78P1	Prenatal 1 Known Mutation incl. MCC	81479
MG78P2	Prenatal 2 Known Mutation incl. MCC	81479
	PCCB - Propionic Acidemia PCC	
L7463	Sanger Sequencing	81406
MG6079	Del-Dup Exonic Microarray	81405
L7507	1 or 2 Known Mutation	81479
MG79P	Prenatal Sequencing incl. MCC	81406
MG79P1	Prenatal 1 Known Mutation incl. MCC	81479
MG79P2	Prenatal 2 Known Mutation incl. MCC	81479
	PDE6C - Achromatopsia	
MG127	Sanger Sequencing	81479
MG6127	Del-Dup Exonic Microarray	81479
MG127C	1 or 2 Known Mutation	81479
MG127P	Prenatal Sequencing incl. MCC	81479
MG127P1	Prenatal 1 Known Mutation incl. MCC	81479
MG127P2	Prenatal 2 Known Mutation incl. MCC	81479
	PDHA1 - Pyurvate Dehydrogenase Complex, PDH	
L7426	Sanger Sequencing	81406
MG6245	Del-Dup Exonic Microarray	81405
MG245C	1 or 2 Known Mutation	81479
MG245P	Prenatal Sequencing incl. MCC	81406
MG245P1	Prenatal 1 Known Mutation incl. MCC	81479
MG245P2	Prenatal 2 Known Mutation incl. MCC	81479
	PKP2 - Arrhythmogenic Right Ventricular Dysplasia (ARVD9)	
MG17	Sanger Sequencing	81406
MG6017	Del-Dup Exonic Microarray	81479
MG17C	1 or 2 Known Mutation	81479
MG17P	Prenatal Sequencing incl. MCC	81406
MG17P1	Prenatal 1 Known Mutation incl. MCC	81479
MG17P2	Prenatal 2 Known Mutation incl. MCC	81479
	PLCB4 - Auricularcondylar Syndrome 2	
MG309	Sanger Sequencing	81479
MG6039	Del-Dup Exonic Microarray	81479
MG309C	1 or 2 Known Mutation	81479

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MG309P	Prenatal Sequencing incl. MCC	81479
MG309P1	Prenatal 1 Known Mutation incl. MCC	81479
MG309P2	Prenatal 2 Known Mutation incl. MCC	81479
	PNPO - Pyridoxal Phosphate Dependent Neonatal Seizures	
L7302	Sanger Sequencing	81479
MG6155	Del-Dup Exonic Microarray	81479
MG155C	1 or 2 Known Mutation	81479
MG155P	Prenatal Sequencing incl. MCC	81479
MG155P1	Prenatal 1 Known Mutation incl. MCC	81479
MG155P2	Prenatal 2 Known Mutation incl. MCC	81479
	POLG1 - POLG1 Related Disorders for Mitochondrial Disorders	
L6911	Sanger Sequencing	81406
L7280	Del-Dup Exonic Microarray	81479
L7281	1 or 2 Known Mutation	81479
MG103P	Prenatal Sequencing incl. MCC	81406
MG103P1	Prenatal 1 Known Mutation incl. MCC	81479
MG103P2	Prenatal 2 Known Mutation incl. MCC	81479

	PTEN - PTEN Related Disorders	
L6942	Sanger Sequencing	81321
L6904	Del-Dup Exonic Microarray	81323
L6910	1 or 2 Known Mutation	81322
MG110P	Prenatal Sequencing incl. MCC	81321
MG110P1	Prenatal 1 Known Mutation incl. MCC	81322
MG110P2	Prenatal 2 Known Mutation incl. MCC	81323
	RB1 - Retinoblastoma	
MG289	Sanger Sequencing	81479
MG6289	Del-Dup Exonic Microarray	81479
MG289C	1 or 2 Known Mutation	81479
MG289P	Prenatal Sequencing incl. MCC	81479
MG289P1	Prenatal 1 Known Mutation incl. MCC	81479
MG289P2	Prenatal 2 Known Mutation incl. MCC	81479
	RET - Multiple Endocrine Neoplasia, Type 2A & 2B	
MG290	Sanger Sequencing	81406
MG6290	Del-Dup Exonic Microarray	81479
MG290C	1 or 2 Known Mutation	81479
MG290P	Prenatal Sequencing incl. MCC	81406
MG290P1	Prenatal 1 Known Mutation incl. MCC	81479
MG290P2	Prenatal 2 Known Mutation incl. MCC	81479
	RNASEH2A - Aicardi-Goutieres Syndrome (AGS)	
MG99	Sanger Sequencing	81479
MG6099	Del-Dup Exonic Microarray	81479
MG99C	1 or 2 Known Mutation	81479
MG99P	Prenatal Sequencing incl. MCC	81479
MG99P1	Prenatal 1 Known Mutation incl. MCC	81479
MG99P2	Prenatal 2 Known Mutation incl. MCC	81479
	RNASEH2B - Aicardi-Goutieres Syndrome (AGS)	
MG97	Sanger Sequencing	81479

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MG6097	Del-Dup Exonic Microarray	81479
MG67C	1 or 2 Known Mutation	81479
MG97P	Prenatal Sequencing incl. MCC	81479
MG97P1	Prenatal 1 Known Mutation incl. MCC	81479
MG97P2	Prenatal 2 Known Mutation incl. MCC	81479
	RNASEH2C - Aicardi-Goutieres Syndrome (AGS)	
MG98	Sanger Sequencing	81479
MG6098	Del-Dup Exonic Microarray	81479
MG98C	1 or 2 Known Mutation	81479
MG98P	Prenatal Sequencing incl. MCC	81479
MG98P1	Prenatal 1 Known Mutation incl. MCC	81479
MG98P2	Prenatal 2 Known Mutation incl. MCC	81479
	RNASET2 - Cystic Leukoencephalopathy	
MG252	Sanger Sequencing	81479
MG6252	Del-Dup Exonic Microarray	81479
MG252C	1 or 2 Known Mutation	81479
MG252P	Prenatal Sequencing incl. MCC	81479
MG252P1	Prenatal 1 Known Mutation incl. MCC	81479
MG252P2	Prenatal 2 Known Mutation incl. MCC	81479

	SALL1 - Townes-Brock Syndrome	
L6920	Sanger Sequencing	81479
L7393	Del-Dup Exonic Microarray	81479
MG48C	1 or 2 Known Mutation	81479
MG48P	Prenatal Sequencing incl. MCC	81479
MG48P1	Prenatal 1 Known Mutation incl. MCC	81479
MG48P2	Prenatal 2 Known Mutation incl. MCC	81479
	SALL4 - Duane Radial Ray Syndrome	
L6932	Sanger Sequencing	81479
L7437	Del-Dup Exonic Microarray	81479
MG49C	1 or 2 Known Mutation	81479
MG49P	Prenatal Sequencing incl. MCC	81479
MG49P1	Prenatal 1 Known Mutation incl. MCC	81479
MG49P2	Prenatal 2 Known Mutation incl. MCC	81479
	SAMHD1 - Aicardi-Goutieres Syndrome (AGS)	
MG123	Sanger Sequencing	81479
MG6123	Del-Dup Exonic Microarray	81479
MG123C	1 or 2 Known Mutation	81479
MG123P	Prenatal Sequencing incl. MCC	81479
MG123P1	Prenatal 1 Known Mutation incl. MCC	81479
MG123P2	Prenatal 2 Known Mutation incl. MCC	81479
	SLC19A3 - Biotin-Responsive Basal Ganglia Disease	
MG308	Sanger Sequencing	81479
MG6308	Del-Dup Exonic Microarray	81479
MG308C	1 or 2 Known Mutation	81479
MG308P	Prenatal Sequencing incl. MCC	81479
MG308P1	Prenatal 1 Known Mutation incl. MCC	81479
MG308P2	Prenatal 2 Known Mutation incl. MCC	81479

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	SLC22A5 - Systemic Primary Carnitine Deficiency	
L7355	Sanger Sequencing	81405
L7356	Del-Dup Exonic Microarray	81479
MG256C	1 or 2 Known Mutation	81479
MG256P	Prenatal Sequencing incl. MCC	81405
MG256P1	Prenatal 1 Known Mutation incl. MCC	81479
MG256P2	Prenatal 2 Known Mutation incl. MCC	81479
	SLC26A4 - Pendred Syndrome	
MG31	Sanger Sequencing	81406
MG6031	Del-Dup Exonic Microarray	81479
MG31C	1 or 2 Known Mutation	81479
MG31P	Prenatal Sequencing incl. MCC	81406
MG31P1	Prenatal 1 Known Mutation incl. MCC	81479
MG31P2	Prenatal 2 Known Mutation incl. MCC	81479
	SLC45A2 - Albinism/Oculocutaneous Albinism: OCA4	
MG91	Sanger Sequencing	81479
MG6091	Del-Dup Exonic Microarray	81479
MG91C	1 or 2 Known Mutation	81479
MG91P	Prenatal Sequencing incl. MCC	81479
MG91P1	Prenatal 1 Known Mutation incl. MCC	81479
MG91P2	Prenatal 2 Known Mutation incl. MCC	81479

	SMAD4 - Hereditary Hemorrhagic Telangiectasia/Juvenile Polyposis Syndrome	
MG291	Sanger Sequencing	81406
MG6291	Del-Dup Exonic Microarray	81405
MG291C	1 or 2 Known Mutation	81479
MG291P	Prenatal Sequencing incl. MCC	81406
MG291P1	Prenatal 1 Known Mutation incl. MCC	81479
MG291P2	Prenatal 2 Known Mutation incl. MCC	81479
	SOX2 - Microopthalmia, Optic Nerve Hypoplasia	
MG258	Sanger Sequencing	81479
MG6258	Del-Dup Exonic Microarray	81479
MG258C	1 or 2 Known Mutation	81479
MG258P	Prenatal Sequencing incl. MCC	81479
MG258P1	Prenatal 1 Known Mutation incl. MCC	81479
MG258P2	Prenatal 2 Known Mutation incl. MCC	81479
	SOX9 - Campomelic Dysplasia with Autosomal Sex Reversal	
L7450	Sanger Sequencing	81479
MG6257	Del-Dup Exonic Microarray	81479
MG257C	1 or 2 Known Mutation	81479
MG257P	Prenatal Sequencing incl. MCC	81479
MG257P1	Prenatal 1 Known Mutation incl. MCC	81479
MG257P2	Prenatal 2 Known Mutation incl. MCC	81479
	SOX10 - Waardenburg Syndrome	
MG35	Sanger Sequencing	81479
MG6035	Del-Dup Exonic Microarray	81479
MG35C	1 or 2 Known Mutation	81479
MG35P	Prenatal Sequencing incl. MCC	81479

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MG35P1	Prenatal 1 Known Mutation incl. MCC	81479
MG35P2	Prenatal 2 Known Mutation incl. MCC	81479
	SPR - Sepiapterin Reductase Deficiency, SPR	
L7349	Sanger Sequencing	81479
L7350	Del-Dup Exonic Microarray	81479
MG80C	1 or 2 Known Mutation	81479
MG80P	Prenatal Sequencing incl. MCC	81479
MG80P1	Prenatal 1 Known Mutation incl. MCC	81479
MG80P2	Prenatal 2 Known Mutation incl. MCC	81479
	SPRED1 - Neurofibromatosis-Like/Legius Syndrome	
MG261	Sanger Sequencing	81405
MG6261	Del-Dup Exonic Microarray	81479
MG261C	1 or 2 Known Mutation	81479
MG261P	Prenatal Sequencing incl. MCC	81405
MG261P1	Prenatal 1 Known Mutation incl. MCC	81479
MG261P2	Prenatal 2 Known Mutation incl. MCC	81479
	SRY - 46, XY Disorder of Sexual Development	
L6916	Sanger Sequencing	81400
L7292	Del-Dup Exonic Microarray	81479
L7362	1 or 2 Known Mutation	81479
MG39P	Prenatal Sequencing incl. MCC	81400
MG39P1	Prenatal 1 Known Mutation incl. MCC	81479
MG39P2	Prenatal 2 Known Mutation incl. MCC	81479

	STK11 - Peutz-Jeghers Syndrome	
L6931	Sanger Sequencing	81405
L7080	Del-Dup Exonic Microarray	81404
MG294C	1 or 2 Known Mutation	81479
MG294P	Prenatal Sequencing incl. MCC	81405
MG294P1	Prenatal 1 Known Mutation incl. MCC	81479
MG294P2	Prenatal 2 Known Mutation incl. MCC	81479
	TBX5 - Holt-Oram Syndrome	
L7049	Sanger Sequencing	81405
L7418	Del-Dup Exonic Microarray	81404
MG50C	1 or 2 Known Mutation	81479
MG50P	Prenatal Sequencing incl. MCC	81405
MG50P1	Prenatal 1 Known Mutation incl. MCC	81479
MG50P2	Prenatal 2 Known Mutation incl. MCC	81479
	TFAP2A - Branchiooculofacial Syndrome	
MG307	Sanger Sequencing	81479
MG6307	Del-Dup Exonic Microarray	81479
MG307C	1 or 2 Known Mutation	81479
MG307P	Prenatal Sequencing incl. MCC	81479
MG307P1	Prenatal 1 Known Mutation incl. MCC	81479
MG307P2	Prenatal 2 Known Mutation incl. MCC	81479
	TREX1 - Aicardi-Goutieres Syndrome (AGS)	
L7043	Sanger Sequencing	81479
MG6096	Del-Dup Exonic Microarray	81479

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L7301	1 or 2 Known Mutation	81479
MG96P	Prenatal Sequencing incl. MCC	81479
MG96P1	Prenatal 1 Known Mutation incl. MCC	81479
MG96P2	Prenatal 2 Known Mutation incl. MCC	81479
	TWIST1 - Saethre-Chotzen Syndrome	
L7040	Sanger Sequencing	81404
L7041	Del-Dup Exonic Microarray	81403
MG259C	1 or 2 Known Mutation	81479
MG259P	Prenatal Sequencing incl. MCC	81404
MG259P1	Prenatal 1 Known Mutation incl. MCC	81479
MG259P2	Prenatal 2 Known Mutation incl. MCC	81479
	TYR - Albinism/Oculocutaneous Albinism: OCA1	
MG87	Sanger Sequencing	81404
MG6087	Del-Dup Exonic Microarray	81403
MG87C	1 or 2 Known Mutation	81479
MG87P	Prenatal Sequencing incl. MCC	81404
MG87P1	Prenatal 1 Known Mutation incl. MCC	81479
MG87P2	Prenatal 2 Known Mutation incl. MCC	81479
	TYRP1 - Albinism/Oculocutaneous Albinism: OCA3	
MG90	Sanger Sequencing, OCA3	81479
MG6090	Del-Dup Exonic Microarray	81479
MG90C	1 or 2 Known Mutation	81479
MG90P	Prenatal Sequencing incl. MCC	81479
MG90P1	Prenatal 1 Known Mutation incl. MCC	81479
MG90P2	Prenatal 2 Known Mutation incl. MCC	81479

	VHL - Von Hippel-Lindau	
L7342	Sanger Sequencing	81404
L7343	Del-Dup Exonic Microarray	81403
MG297C	1 or 2 Known Mutation	81479
MG297P	Prenatal Sequencing incl. MCC	81404
MG297P1	Prenatal 1 Known Mutation incl. MCC	81479
MG297P2	Prenatal 2 Known Mutation incl. MCC	81479
	WFS1 - Wolfram Syndrome	
MG137	Sanger Sequencing, OCA3	81479
MG6137	Del-Dup Exonic Microarray	81479
MG137C	1 or 2 Known Mutation	81479
MG137P	Prenatal Sequencing incl. MCC	81479
MG137P1	Prenatal 1 Known Mutation incl. MCC	81479
MG137P2	Prenatal 2 Known Mutation incl. MCC	81479
	WT1 - 46, XY Disorder of Sexual Development/Fraser Syndrome/Denys-Drash Syndrome	
L6727	Sanger Sequencing	81405
L7275	Del-Dup Exonic Microarray	81479
L7261	1 or 2 Known Mutation	81479
L6728	Prenatal Sequencing incl. MCC	81405
MG40P1	Prenatal 1 Known Mutation incl. MCC	81479
MG40P2	Prenatal 2 Known Mutation incl. MCC	81479

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