

Panels Offered		
Test Code	Panel - Test Description	CPT Codes
L7464 MG6052	3-Methyl-Crotonyl-CoA Carboxylase Deficiency - 3MCC A+B 2 Gene Sanger Sequencing (MCCC1 & MCCC2) 2 Gene Del-Dup Exonic Microarray (MCCC1 & MCCC2)	81406x2 81479x2
L7332	46, XY Disorder of Sexual Development 3 Gene Sanger Sequencing (AR, SRY, WT1)	81400 & 81405x2
MG93 L6921	Aicardi-Goutieres Syndrome (AGS) 5 Gene Sanger Sequencing (TREX1, RNASEH2B, RNASEH2C, RNASEH2A, SAMHD1) 5 Gene Del-Dup Exonic Microarray (TREX1, RNASEH2B, RNASEH2C, RNASEH2A, SAMHD1)	81479x5 81479x5
L6723 L6724	Albinism/Oculocutaneous Albinism: OA1, OCA1, OCA2 and OCA2 deletion, OCA3, OCA4 5 Gene Sanger Sequencing (OA1, OCA1, OCA2, OCA3, OCA4) 5 Gene Del-Dup Exonic Microarray (OA1, OCA1, OCA2, OCA3, OCA4)	81404 & 81479x4 81479x5
MG16 MG6016 MG16C	Arrhythmogenic Right Ventricular Dysplasia 4 Gene Sanger Sequencing (PKP2, DSP, DSG2, DSC2) 4 Gene Del-Dup Exonic Microarray (PKP2, DSP, DSG2, DSC2) 1 Known Mutation in the 4 ARVD	81406x4 81406x4 81479
MG284 MG6284	Chronic Granulomatous Disease 3 Gene Sanger Sequencing (CYBA, CYBB, NCF2) 3 Gene Del-dup Exonic Microarray (CYBA, CYBB, NCF2)	81479x3 81479x3
L7039 L6926 MG241P	Fibroblast Growth Factor Receptor Disorders - FGFR 3 Gene Sanger Sequencing (FGFR1, FGFR2, FGFR3) 3 Gene Del-Dup Exonic Microarray (FGFR1, FGFR2, FGFR3) 3 Gene Prenatal Sanger Sequencing incl. MCC (FGFR1, FGFR2, FGFR3)	81479x3 81479x3 81479x3 & 81265 or 81266*
L6936 MG6057	Glutaric Acidemia Type 2 - GA2 3 Gene Sanger Sequencing (ETFA, ETFB, ETFDH) 3 Gene Del-Dup Exonic Microarray (ETFA, ETFB, ETFDH)	81479x3 81479x3
L7052 MG66 MG6066	Methylmalonic Acidemia - MMA 24 Gene NextGen Sequencing Panel 3 Gene Sanger Sequencing (MMAA, MMAB, MUT) 3 Gene Del-Dup Exonic Microarray (MMAA, MMAB, MUT)	81401, 81406x3, 81405x2, 81479x18 81479x3 81479x3
L6928 L7152	Non-Ketotic Hyperglycinemia (Glycine Encephalopathy) - NKH 3 Gene Sanger Sequencing (AMT, GCSH, GLDC) 3 Gene Del-Dup Exonic Microarray (AMT, GCSH, GLDC)	81479x3 81479x3
MG77 MG6077	Propionic Acidemia - PCC 2 Gene Sanger Sequencing (PCCA, PCCB) 2 Gene Del-Dup Exonic Microarray (PCCA, PCCB)	81406x2 81405 & 81479
L7273 MG6292 L7424 MG292P MG292P1 MG292P2	RASopathy Spectrum Disorders (Noonan) NextGen Sequencing (NGS) Single Gene Del-Dup OGT Microarray 1 or 2 Known Mutation Prenatal NGS incl. MCC Prenatal 1 Known Mutation incl. MCC Prenatal 2 Known Mutation incl. MCC	81442 81323 81322 81442 & 81265 or 81266* 81322 & 81265 or 81266* 81323 & 81265 or 81266*
L7124 L7125	Rubinstein-Taybi Syndrome 2 Gene Sanger Sequencing (CREBBP, EP300) 2 Gene Del-Dup Exonic Microarray (CREBBP, EP300)	81407 & 81479 81406 & 81479
MG32	Waardenburg Syndrome 4 Gene Sanger Sequencing (EDNRB, MITF, PAX3, SOX10)	81479x4

Individual Genes Offered		
Test Code	Gene - Test Description	CPT
MG124 MG6124 MG124C MG124P MG124P1 MG124P2	ABCA4 - Stargardt, arRP, Age-Related Macular Degeneration, CORD3 Sanger Sequencing Del-Dup Exonic Microarray 1 or 2 Known Mutation Prenatal Sequencing incl. MCC Prenatal 1 Known Mutation incl. MCC Prenatal 2 Known Mutation incl. MCC	81408 81479 81479 81408 & 81265 or 81266* 81479 & 81265 or 81266* 81479 & 81265 or 81266*
L6917 L7279 L7277 L7278 MG65P MG65P1 MG65P2	ACADM - Medium Chain Acyl-CoA Dehydrogenase Deficiency, MCAD Sanger Sequencing Sanger Sequencing Common Mutation (K304E) Del-Dup Exonic Microarray 1 or 2 Known Mutation Prenatal Sequencing incl. MCC Prenatal 1 Known Mutation incl. MCC Prenatal 2 Known Mutation incl. MCC	81479 81400 81479 81479 81479 & 81265 or 81266* 81479 & 81265 or 81266* 81479 & 81265 or 81266*

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L6906	ACADVL - Very Long Chain Acyl-CoA Dehydrogenase Deficiency, VLCAD Sanger Sequencing Del-Dup Exonic Microarray 1 or 2 Known Mutation Prenatal Sequencing incl. MCC Prenatal 1 Known Mutation incl. MCC Prenatal 2 Known Mutation incl. MCC	81406
L7161		81479
L7206		81403
MGP		81406 & 81265 or 81266*
MGP1		81479 & 81265 or 81266*
MGP2		81479 & 81265 or 81266*
MG262	ACVRL1/ALK1 - Hereditary Hemorrhagic Telangiectasia Sanger Sequencing Del-Dup Exonic Microarray 1 or 2 Known Mutation Prenatal Sequencing incl. MCC Prenatal 1 Known Mutation incl. MCC Prenatal 2 Known Mutation incl. MCC	81479
MG6262		81479
MG262C		81479
MG262P		81479 & 81265 or 81266*
MG262P1		81479 & 81265 or 81266*
MG262P2		81479 & 81265 or 81266*
MG55	ALDH7A1 - Antiquitin, Pyridoxine-Dependent Neonatal Seizures, ATQ-1 Sanger Sequencing Del-Dup Exonic Microarray 1 or 2 Known Mutation Prenatal Sequencing incl. MCC Prenatal 1 Known Mutation incl. MCC Prenatal 2 Known Mutation incl. MCC	81406
MG6055		81479
MG55C		81479
MG55P		81406 & 81265 or 81266*
MG55P1		81479 & 81265 or 81266*
MG55P2		81479 & 81265 or 81266*
MG347	ALX4 - Frontonasal Dysplasia 2; Parietal Foramina 2 Sanger Sequencing Del-Dup Exonic Microarray 1 or 2 Known Mutation Prenatal Sequencing incl. MCC Prenatal 1 Known Mutation incl. MCC Prenatal 2 Known Mutation incl. MCC	81479
MG6347		81479
MG347C		81479
MG347P		81479 & 81265 or 81266*
MG347P1		81479 & 81265 or 81266*
MG347P2		81479 & 81265 or 81266*
L7153	AMT - Non-Ketotic Hyperglycinemia, Glycine Encephalopathy, NKH Sanger Sequencing Del-Dup Exonic Microarray 1 or 2 Known Mutation Prenatal Sequencing incl. MCC Prenatal 1 Known Mutation incl. MCC Prenatal 2 Known Mutation incl. MCC	81479
L7154		81479
L7207		81479
MG74P		81479 & 81265 or 81266*
MG74P1		81479 & 81265 or 81266*
MG74P2		81479 & 81265 or 81266*
MG267	APC - Familial adenomatous polyposis (FAP)/Gardner syndrome/Turcot Syndrome Sanger Sequencing Del-Dup Exonic Microarray 1 or 2 Known Mutation Prenatal Sequencing incl. MCC Prenatal 1 Known Mutation incl. MCC Prenatal 2 Known Mutation incl. MCC	81201
MG6267		81203
MG267C		81202
MG267P		81201 & 81265 or 81266*
MG267P1		81202 & 81265 or 81266*
MG267P2		81202 & 81265 or 81266*
L7174	AR - 46, XY Disorder of Sexual Development Sanger Sequencing Del-Dup Exonic Microarray 1 or 2 Known Mutation Prenatal Sequencing incl. MCC Prenatal 1 Known Mutation incl. MCC Prenatal 2 Known Mutation incl. MCC	81405
L7290		81479
L7291		81479
MG38P		81405 & 81265 or 81266*
MG38P1		81479 & 81265 or 81266*
MG38P2		81479 & 81265 or 81266*
MG348	BAG3 - Dilated Cardiomyopathy; Myofibrillar Myopathy; Primary Dilated Cardiomyopathy Sanger Sequencing 1 or 2 Known Mutation Prenatal Sequencing incl. MCC Prenatal 1 Known Mutation incl. MCC Prenatal 2 Known Mutation incl. MCC	81479
MG348C		81479
MG348P		81479 & 81265 or 81266*
MG348P1		81479 & 81265 or 81266*
MG348P2		81479 & 81265 or 81266*
L7268		BTD - Biotinidase Deficiency Sanger Sequencing Del-Dup Exonic Microarray 1 or 2 Known Mutation Prenatal Sequencing incl. MCC Prenatal 1 Known Mutation incl. MCC Prenatal 2 Known Mutation incl. MCC
L7359	81479	
MG126C	81479	
MG126P	81404 & 81265 or 81266*	
MG126P1	81479 & 81265 or 81266*	
MG126P2	81479 & 81265 or 81266*	
MG100	BCKDHA Y438N & BCKDHB R183P, G278S & E372X - Maple Syrup Disease - Common Jewish Mutations Sanger Sequencing 1 or 2 Known Mutation	81479
MG100C		81479

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MG243	BOLA3 - Multiple Mitochondrial Dysfunctions Syndrome, MMDS Sanger Sequencing	81479
MG6243	Del-Dup Exonic Microarray	81479
MG243C	1 or 2 Known Mutation	81479
MG243P	Prenatal Sequencing incl. MCC	81479 & 81265 or 81266*
MG243P1	Prenatal 1 Known Mutation incl. MCC	81479 & 81265 or 81266*
MG243P2	Prenatal 2 Known Mutation incl. MCC	81479 & 81265 or 81266*
MG105	C-KIT - Piebaldism, Mast Cell Leukemia, GIST Sanger Sequencing	81479
MG6105	Del-Dup Exonic Microarray	81479
MG105C	1 or 2 Known Mutation	81479
MG105P	Prenatal Sequencing incl. MCC	81479 & 81265 or 81266*
MG105P1	Prenatal 1 Known Mutation incl. MCC	81479 & 81265 or 81266*
MG105P2	Prenatal 2 Known Mutation incl. MCC	81479 & 81265 or 81266*
L7300	C7orf10 - Glutaric Acidemia Type 3, GA3 Sanger Sequencing	81479
L7483	Del-Dup Exonic Microarray	81479
MG102C	1 or 2 Known Mutation	81479
MG102P	Prenatal Sequencing incl. MCC	81479 & 81265 or 81266*
MG102P1	Prenatal 1 Known Mutation incl. MCC	81479 & 81265 or 81266*
MG102P2	Prenatal 2 Known Mutation incl. MCC	81479 & 81265 or 81266*
L7346	CBS - Homocystinuria due to Cystathionine Beta-Synthase Deficiency Sanger Sequencing	81406
L7286	Del-Dup Exonic Microarray	81479
L7287	1 or 2 Known Mutation	81479
MG62P	Prenatal Sequencing incl. MCC	81406 & 81265 or 81266*
MG62P1	Prenatal 1 Known Mutation incl. MCC	81479 & 81265 or 81266*
MG62P2	Prenatal 2 Known Mutation incl. MCC	81479 & 81265 or 81266*
MG260	CDKL5 - Atypical Rett Syndrome Sanger Sequencing	81406
MG6260	Del-Dup Exonic Microarray	81405
MG260C	1 or 2 Known Mutation	81479
MG260P	Prenatal Sequencing incl. MCC	81406 & 81265 or 81266*
MG260P1	Prenatal 1 Known Mutation incl. MCC	81479 & 81265 or 81266*
MG261P2	Prenatal 2 Known Mutation incl. MCC	81479 & 81265 or 81266*
MG107	CEP290 - Joubert Syndrome, Bardet-Biedl, Meckel, Senior-Loken, LCA Sanger Sequencing	81408
MG6107	Del-Dup Exonic Microarray	81479
MG107C	1 or 2 Known Mutation	81479
MG107P	Prenatal Sequencing incl. MCC	81408 & 81265 or 81266*
MG107P1	Prenatal 1 Known Mutation incl. MCC	81479 & 81265 or 81266*
MG107P2	Prenatal 2 Known Mutation incl. MCC	81479 & 81265 or 81266*
L7050	CFTR - Cystic Fibrosis 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 & 81265 or 81266*
L6913	CHD7 - Charge Syndrome Sanger Sequencing	81407
L7324	Del-Dup Exonic Microarray	81479
MG272C	1 or 2 Known Mutation	81479
MG272P	Prenatal Sequencing incl. MCC	81407 & 81265 or 81266*
MG272P1	Prenatal 1 Known Mutation incl. MCC	81479 & 81265 or 81266*
MG272P2	Prenatal 2 Known Mutation incl. MCC	81479 & 81265 or 81266*
MG83	CHS1 (LYST) - Chediak-Higashi Syndrome Sanger Sequencing	81479
MG6083	Del-Dup Exonic Microarray	81479
MG83C	1 or 2 Known Mutation	81479
MG83P	Prenatal Sequencing incl. MCC	81479 & 81265 or 81266*
MG83P1	Prenatal 1 Known Mutation incl. MCC	81479 & 81265 or 81266*
MG83P2	Prenatal 2 Known Mutation incl. MCC	81479 & 81265 or 81266*
MG119	CNGA3 - Achromatopsia Sanger Sequencing	81479
MG6119	Del-Dup Exonic Microarray	81479
MG119C	1 or 2 Known Mutation	81479
MG119P	Prenatal Sequencing incl. MCC	81479 & 81265 or 81266*
MG119P1	Prenatal 1 Known Mutation incl. MCC	81479 & 81265 or 81266*
MG119P2	Prenatal 2 Known Mutation incl. MCC	81479 & 81265 or 81266*

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MG120	CNGB3 - Achromatopsia Sanger Sequencing	81479
MG6120	Del-Dup Exonic Microarray	81479
MG120C	1 or 2 Known Mutation	81479
MG120P	Prenatal Sequencing incl. MCC	81479 & 81265 or 81266*
MG120P1	Prenatal 1 Known Mutation incl. MCC	81479 & 81265 or 81266*
MG120P2	Prenatal 2 Known Mutation incl. MCC	81479 & 81265 or 81266*
L6908	CREBBP - Rubinstein-Taybi Syndrome Sanger Sequencing	81407
L7271	Del-Dup Exonic Microarray	81406
L7427	1 or 2 Known Mutation	81479
MG112P	Prenatal Sequencing incl. MCC	81407 & 81265 or 81266*
MG112P1	Prenatal 1 Known Mutation incl. MCC	81479 & 81265 or 81266*
MG112P2	Prenatal 2 Known Mutation incl. MCC	81479 & 81265 or 81266*
MG152	CYBA - Chronic Granulomatous Disease Sanger Sequencing, p22-PHOX	81479
MG6152	Del-Dup Exonic Microarray	81479
MG152C	1 or 2 Known Mutation	81479
MG152P	Prenatal Sequencing incl. MCC	81479 & 81265 or 81266*
MG152P1	Prenatal 1 Known Mutation incl. MCC	81479 & 81265 or 81266*
MG152P2	Prenatal 2 Known Mutation incl. MCC	81479 & 81265 or 81266*
L6922	CYBB - Chronic Granulomatous Disease Sanger Sequencing, p91-PHOX	81479
L7340	Del-Dup Exonic Microarray	81479
L7428	1 or 2 Known Mutation	81479
MG151P	Prenatal Sequencing incl. MCC	81479 & 81265 or 81266*
MG151P1	Prenatal 1 Known Mutation incl. MCC	81479 & 81265 or 81266*
MG151P2	Prenatal 2 Known Mutation incl. MCC	81479 & 81265 or 81266*
MG20	DSC2 - Arrhythmogenic Right Ventricular Dysplasia (ARVD11) Sanger Sequencing	81406
MG6020	Del-Dup Exonic Microarray	81479
MG20C	1 or 2 Known Mutation	81479
MG20P	Prenatal Sequencing incl. MCC	81406 & 81265 or 81266*
MG20P1	Prenatal 1 Known Mutation incl. MCC	81479 & 81265 or 81266*
MG20P2	Prenatal 2 Known Mutation incl. MCC	81479 & 81265 or 81266*
MG20	DSG2 - Arrhythmogenic Right Ventricular Dysplasia (ARVD10) Sanger Sequencing	81406
MG6020	Del-Dup Exonic Microarray	81479
MG20C	1 or 2 Known Mutation	81479
MG20P	Prenatal Sequencing incl. MCC	81406 & 81265 or 81266*
MG20P1	Prenatal 1 Known Mutation incl. MCC	81479 & 81265 or 81266*
MG20P2	Prenatal 2 Known Mutation incl. MCC	81479 & 81265 or 81266*
MG19	DSP- Arrhythmogenic Right Ventricular Dysplasia (ARVD8) Sanger Sequencing	81406
MG6019	Del-Dup Exonic Microarray	81479
MG19C	1 or 2 Known Mutation	81479
MG19P	Prenatal Sequencing incl. MCC	81406 & 81265 or 81266*
MG19P1	Prenatal 1 Known Mutation incl. MCC	81479 & 81265 or 81266*
MG19P2	Prenatal 2 Known Mutation incl. MCC	81479 & 81265 or 81266*
L6909	DHCR7 - Smith Lemli Opitz, SLOS Sanger Sequencing	81405
L7288	Del-Dup Exonic Microarray	81479
L7289	1 or 2 Known Mutation	81479
MG136P	Prenatal Sequencing incl. MCC	81405 & 81265 or 81266*
MG136P1	Prenatal 1 Known Mutation incl. MCC	81479 & 81265 or 81266*
MG136P2	Prenatal 2 Known Mutation incl. MCC	81479 & 81265 or 81266*
MG349	DHH - 46, XY Disorder of Sexual Development Sanger Sequencing	81479
MG6349	Del-Dup Exonic Microarray	81479
MG349C	1 or 2 Known Mutation	81479
MG349P	Prenatal Sequencing incl. MCC	81479 & 81265 or 81266*
MG349P1	Prenatal 1 Known Mutation incl. MCC	81479 & 81265 or 81266*
MG349P2	Prenatal 2 Known Mutation incl. MCC	81479 & 81265 or 81266*
MG254	EDN3 - Waardenburg Syndrome Sanger Sequencing	81479
MG6254	Del-Dup Exonic Microarray	81479
MG254C	1 or 2 Known Mutation	81479
MG254P	Prenatal Sequencing incl. MCC	81479 & 81265 or 81266*
MG254P1	Prenatal 1 Known Mutation incl. MCC	81479 & 81265 or 81266*
MG254P2	Prenatal 2 Known Mutation incl. MCC	81479 & 81265 or 81266*

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MG36	EDNRB - Waardenburg Syndrome Sanger Sequencing	81479
MG6036	Del-Dup Exonic Microarray	81479
MG36C	1 or 2 Known Mutation	81479
MG36P	Prenatal Sequencing incl. MCC	81479 & 81265 or 81266*
MG36P1	Prenatal 1 Known Mutation incl. MCC	81479 & 81265 or 81266*
MG36P2	Prenatal 2 Known Mutation incl. MCC	81479 & 81265 or 81266*
L6923	EFNB1 - Craniofrontonasal Dysplasia Sanger Sequencing	81479
MG6253	Del-Dup Exonic Microarray	81479
MG253C	1 or 2 Known Mutation	81479
MG253P	Prenatal Sequencing incl. MCC	81479 & 81265 or 81266*
MG253P1	Prenatal 1 Known Mutation incl. MCC	81479 & 81265 or 81266*
MG253P2	Prenatal 2 Known Mutation incl. MCC	81479 & 81265 or 81266*
L7325	EFTUD2 - Mandibulofacial Dysostosis with Microcephaly Sanger Sequencing	81479
L7384	Del-Dup Exonic Microarray	81479
L7386	1 or 2 Known Mutation	81479
MG304P	Prenatal Sequencing incl. MCC	81479 & 81265 or 81266*
MG304P1	Prenatal 1 Known Mutation incl. MCC	81479 & 81265 or 81266*
MG304P2	Prenatal 2 Known Mutation incl. MCC	81479 & 81265 or 81266*
MG306	ELANE - Severe Congenital Neutropenia and Cyclic Neutropenia Sanger Sequencing	81479
MG6306	Del-Dup Exonic Microarray	81479
MG306C	1 or 2 Known Mutation	81479
MG306P	Prenatal Sequencing incl. MCC	81479 & 81265 or 81266*
MG306P1	Prenatal 1 Known Mutation incl. MCC	81479 & 81265 or 81266*
MG306P2	Prenatal 2 Known Mutation incl. MCC	81479 & 81265 or 81266*
MG263	ENG - Hereditary Hemorrhagic Telangiectasia Sanger Sequencing	81406
MG6263	Del-Dup Exonic Microarray	81405
MG263C	1 or 2 Known Mutation	81479
MG263P	Prenatal Sequencing incl. MCC	81406 & 81265 or 81266*
MG263P1	Prenatal 1 Known Mutation incl. MCC	81479 & 81265 or 81266*
MG263P2	Prenatal 2 Known Mutation incl. MCC	81479 & 81265 or 81266*
L7267	EP300 - Rubenstein-Taybi Syndrome Sanger Sequencing	81479
L7266	Del-Dup Exonic Microarray	81479
L7341	1 or 2 Known Mutation	81479
MG113P	Prenatal Sequencing incl. MCC	81479 & 81265 or 81266*
MG113P1	Prenatal 1 Known Mutation incl. MCC	81479 & 81265 or 81266*
MG113P2	Prenatal 2 Known Mutation incl. MCC	81479 & 81265 or 81266*
L7305	ETFA - Glutaric Acidemia Type 2, GA2 Sanger Sequencing	81479
L7306	Del-Dup Exonic Microarray	81479
L7307	1 or 2 Known Mutation	81479
MG59P	Prenatal Sequencing incl. MCC	81479 & 81265 or 81266*
MG59P1	Prenatal 1 Known Mutation incl. MCC	81479 & 81265 or 81266*
MG59P2	Prenatal 2 Known Mutation incl. MCC	81479 & 81265 or 81266*
L7308	ETFB - Glutaric Acidemia Type 2, GA2 Sanger Sequencing	81479
L7309	Del-Dup Exonic Microarray	81479
L7310	1 or 2 Known Mutation	81479
MG60P	Prenatal Sequencing incl. MCC	81479 & 81265 or 81266*
MG60P1	Prenatal 1 Known Mutation incl. MCC	81479 & 81265 or 81266*
MG60P2	Prenatal 2 Known Mutation incl. MCC	81479 & 81265 or 81266*
L7311	ETFDH - Glutaric Acidemia Type 2, GA2 Sanger Sequencing	81479
L7312	Del-Dup Exonic Microarray	81479
L7313	1 or 2 Known Mutation	81479
MG58P	Prenatal Sequencing incl. MCC	81479 & 81265 or 81266*
MG58P1	Prenatal 1 Known Mutation incl. MCC	81479 & 81265 or 81266*
MG58P2	Prenatal 2 Known Mutation incl. MCC	81479 & 81265 or 81266*
L7262	FGFR1 - Pfeiffer Syndrome, Osteoglophonic Dysplasia 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 & 81265 or 81266*
MG128P1	Prenatal 1 Known Mutation incl. MCC	81479 & 81265 or 81266*
MG128P2	Prenatal 2 Known Mutation incl. MCC	81479 & 81265 or 81266*

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L7046 L7282 L7283 MG129P MG129P1 MG129P2	FGFR2 - Crouzon, Jackson-Weiss, Beare-Stevenson, Apert, Pfeiffer Sanger Sequencing Del-Dup Exonic Microarray 1 or 2 Known Mutation Prenatal Sequencing incl. MCC Prenatal 1 Known Mutation incl. MCC Prenatal 2 Known Mutation incl. MCC	81404 81479 81479 81404 & 81265 or 81266* 81479 & 81265 or 81266* 81479 & 81265 or 81266*
L7048 MG236P1 MG236P2	FGFR2 - Apert Specific Sanger Sequencing for S252W, P253R Mutations Prenatal 1 Known Mutation incl. MCC Prenatal 2 Known Mutation incl. MCC	81404 81404 & 81265 or 81266* 81404 & 81265 or 81266*
L7344 MG237P1 MG237P2	FGFR2 - Beare-Stevenson Specific Sanger Sequencing for S372C, Y375C Mutations Prenatal 1 Known Mutation incl. MCC Prenatal 2 Known Mutation incl. MCC	81404 81404 & 81265 or 81266* 81479 & 81265 or 81266*
L6907 L7272 L7263 MG130P MG130P1 MG130P2 L7299 LF1054 L7264 MG142P	FGFR3 - Achondroplasia, Hypochondroplasia, Thanaphoric Dysplasia, SADDAN, Craniosyostoses, Crouzon, Muenke Sanger Sequencing Del-Dup Exonic Microarray 1 or 2 Known Mutation Prenatal Sequencing incl. MCC Prenatal 1 Known Mutation incl. MCC Prenatal 2 Known Mutation incl. MCC Tier 1 (sanger sequencing exons 7, 9, 12, 14 & 18) Prenatal Tier 1 incl. MCC Add-On Sequencing <i>after</i> Tier 1 Prenatal Add-On Sequencing <i>after</i> Tier 1 incl. MCC	81479 81479 81403 81479 & 81265 or 81266* 81479 & 81265 or 81266* 81479 & 81265 or 81266* 81403, 81404 & 81265 or 81266* 81404, 81479 & 81265 or 81266* 81479 81479 & 81265 or 81266*
L7338 MG44P	FGFR3 - Crouzon (A391E) Syndrome Specific Sanger Sequencing Prenatal Sequencing incl. MCC	81404 81404 & 81265 or 81266*
L6914 MG43P	FGFR3 - Muenke (P250R Variant) Syndrome Specific Sanger Sequencing Prenatal Sequencing incl. MCC	81400 81400 & 81265 or 81266*
L7304 MG45P	FGFR3 - Saddam (K650M) Specific Sanger Sequencing Prenatal Sequencing incl. MCC	81404 81404 & 81265 or 81266*
L6927 L7284 L7285	FMO3 - Trimethylaminuria, TMA Sanger Sequencing Del-Dup Exonic Microarray 1 or 2 Known Mutation	81479 81479 81479
L6943 L7204 MG46P	FMR1 - Fragile X: FX CGG repeats, FX (includes Southern Blot if positive) Del-Dup Exonic Microarray Prenatal CGG Repeats, FX incl. MCC (includes Southern Blot if positive)	81243 & 81244 81479 81243, 81244 & 81265 or 81266*
L6905 L7160 L7205 MG56P MG56P1 MG56P2	GCDH - Glutaric Acidemia Type 1, GA1 Sanger Sequencing Del-Dup Exonic Microarray 1 or 2 Known Mutation Prenatal Sequencing incl. MCC Prenatal 1 Known Mutation incl. MCC Prenatal 2 Known Mutation incl. MCC	81406 81479 81403 81406 & 81265 or 81266* 81403 & 81265 or 81266* 81403 & 81265 or 81266*
L7157 L7158 L7208 MG76P MG76P1 MG76P2	GCSH - Non-Ketotic Hyperglycinemia, Glycine Encephalopathy, NKH Sanger Sequencing Del-Dup Exonic Microarray 1 or 2 Known Mutation Prenatal Sequencing incl. MCC Prenatal 1 Known Mutation incl. MCC Prenatal 2 Known Mutation incl. MCC	81479 81479 81479 81479 & 81265 or 81266* 81479 & 81265 or 81266* 81479 & 81265 or 81266*
L6919 MG6029 MG29C MG29P MG29P1 MG29P2	GJB2 - Connexin 26 - Related Hearing Loss Sanger Sequencing Del-Dup Exonic Microarray 1 or 2 Known Mutation Prenatal Sequencing incl. MCC Prenatal 1 Known Mutation incl. MCC Prenatal 2 Known Mutation incl. MCC	81252 81479 81253 81252 & 81265 or 81266* 81253 & 81265 or 81266* 81253 & 81265 or 81266*
MG30 MG6030 MG30P1	GJB6 - Connexin 30 - Related Hearing Loss Common Deletion Del-Dup Exonic Microarray Common Deletion Prenatal 1 Known Mutation incl. MCC	81254 81479 81254 & 81265 or 81266*

*81266 will be applied for twin specimens

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L7155	GLDC - Non-Ketotic Hyperglycinemia, Glycine Encephalopathy, NKH Sanger Sequencing	81479
L7156	Del-Dup Exonic Microarray	81479
L6726	1 or 2 Known Mutation	81479
L7328	Prenatal Sequencing incl. MCC	81479 & 81265 or 81266*
L7327	Prenatal 1 Known Mutation incl. MCC	81479 & 81265 or 81266*
L7326	Prenatal 2 Known Mutation incl. MCC	81479 & 81265 or 81266*
L7171	GNAI3 - Auriculocondylar Syndrome Type 1 Sanger Sequencing	81479
L7170	Del-Dup Exonic Microarray	81479
MG305C	1 or 2 Known Mutation	81479
MG305P	Prenatal Sequencing incl. MCC	81479 & 81265 or 81266*
MG305P1	Prenatal 1 Known Mutation incl. MCC	81479 & 81265 or 81266*
MG305P2	Prenatal 2 Known Mutation incl. MCC	81479 & 81265 or 81266*
MG121	GNAT2 - Achromatopsia Sanger Sequencing	81479
MG6121	Del-Dup Exonic Microarray	81479
MG121C	1 or 2 Known Mutation	81479
MG121P	Prenatal Sequencing incl. MCC	81479 & 81265 or 81266*
MG121P1	Prenatal 1 Known Mutation incl. MCC	81479 & 81265 or 81266*
MG121P2	Prenatal 2 Known Mutation incl. MCC	81479 & 81265 or 81266*
MG92	GPR143 - Albinism/Oculocutaneous Albinism: OA1 (X-linked) Sanger Sequencing	81479
MG6092	Del-Dup Exonic Microarray	81479
MG92C	1 or 2 Known Mutation	81479
MG92P	Prenatal Sequencing incl. MCC	81479 & 81265 or 81266*
MG92P1	Prenatal 1 Known Mutation incl. MCC	81479 & 81265 or 81266*
MG92P2	Prenatal 2 Known Mutation incl. MCC	81479 & 81265 or 81266*
L6933	HADHA - Long Chain 3-Hydroxy-Acyl-CoA Dehydrogenase Deficiency, LCHAD Sanger Sequencing of Common Mutation (E5100Q)	81479
L7276	Del-Dup Exonic Microarray	81479
MG63P	Prenatal Sequencing of E5100Q incl. MCC	81479 & 81265 or 81266*
MG63P1	Prenatal 1 Known Mutation of E5100Q incl. MCC	81479 & 81265 or 81266*
L6930	HFE - Hereditary Hemochromatosis Sanger Sequencing of exons 2 & 4 to detect C282Y, H63D mutations	81256
MG61	HLCS - Holocarboxylase Synthetase Deficiency Sanger Sequencing	81406
MG6061	Del-Dup Exonic Microarray	81479
MG61C	1 or 2 Known Mutation	81479
MG61P	Prenatal Sequencing incl. MCC	81406 & 81265 or 81266*
MG61P1	Prenatal 1 Known Mutation incl. MCC	81479 & 81265 or 81266*
MG61P2	Prenatal 2 Known Mutation incl. MCC	81479 & 81265 or 81266*
MG84	HPS1 - Hermansky - Pudlak Syndrome: Common Puerto Rican Mutation 1470_1486dup16 Sanger Sequencing	81779
MG84P	Prenatal Sequencing incl. MCC	81479 & 81265 or 81266*
MG85	HPS3 - Hermansky - Pudlak Syndrome: Common Puerto Rican Mutation 339_4260del3904 Sanger Sequencing	81479
MG85P	Prenatal Sequencing incl. MCC	81479 & 81265 or 81266*
MG86	HPS3 - Hermansky - Pudlak Syndrome: Common Ashkenazi Jewish Mutation 1163+1G>A Sanger Sequencing	81479
MG86P	Prenatal Sequencing incl. MCC	81479 & 81265 or 81266*
L7469	L1CAM - Hydrocephalus Due to Congenital Stenosis of Aqueduct of Sylvius/MASA Syndrome Sanger Sequencing	81407
MG6282	Del-Dup Exonic Microarray	81479
MG282C	1 or 2 Known Mutation	81479
MG282P	Prenatal Sequencing incl. MCC	81407 & 81265 or 81266*
MG282P1	Prenatal 1 Known Mutation incl. MCC	81479 & 81265 or 81266*
MG282P2	Prenatal 2 Known Mutation incl. MCC	81479 & 81265 or 81266*
MG22	LAMP2 - Danon Disease, GSD IIB (X-linked) Sanger Sequencing	81405
MG6022	Del-Dup Exonic Microarray	81479
MG22C	1 or 2 Known Mutation	81479
MG22P	Prenatal Sequencing incl. MCC	81405 & 81265 or 81266*
MG22P1	Prenatal 1 Known Mutation incl. MCC	81479 & 81265 or 81266*
MG22P2	Prenatal 2 Known Mutation incl. MCC	81479 & 81265 or 81266*
MG283	LIAS - Pyruvate Dehydrogenase Lipoic Acid Synthetase Deficiency Sanger Sequencing	81479
MG283C	1 or 2 Known Mutation	81479
MG283P	Prenatal Sequencing incl. MCC	81479 & 81265 or 81266*
MG283P1	Prenatal 1 Known Mutation incl. MCC	81479 & 81265 or 81266*
MG283P2	Prenatal 2 Known Mutation incl. MCC	81479 & 81265 or 81266*

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MG242	LMBRD1 - Cobalamin F Deficiency Sanger Sequencing	81479
MG6242	Del-Dup Exonic Microarray	81479
MG242C	1 or 2 Known Mutation	81479
MG242P	Prenatal Sequencing incl. MCC	81479 & 81265 or 81266*
MG242P1	Prenatal 1 Known Mutation incl. MCC	81479 & 81265 or 81266*
MG242P2	Prenatal 2 Known Mutation incl. MCC	81479 & 81265 or 81266*
MG23	LMNA - Lamin A/C Sanger Sequencing	81406
MG6023	Del-Dup Exonic Microarray	81479
L7339	1 or 2 Known Mutation	81479
MG23P	Prenatal Sequencing incl. MCC	81406 & 81265 or 81266*
MG23P1	Prenatal 1 Known Mutation incl. MCC	81479 & 81265 or 81266*
MG23P2	Prenatal 2 Known Mutation incl. MCC	81479 & 81265 or 81266*
MG53	MCCCI - 3-Methyl-Crotonyl-CoA Carboxylase Deficiency Sanger Sequencing	81406
MG6053	Del-Dup Exonic Microarray	81479
MG53C	1 or 2 Known Mutation	81479
MG53P	Prenatal Sequencing incl. MCC	81406 & 81265 or 81266*
MG53P1	Prenatal 1 Known Mutation incl. MCC	81479 & 81265 or 81266*
MG53P2	Prenatal 2 Known Mutation incl. MCC	81479 & 81265 or 81266*
MG54	MCCC2 - 3-Methyl-Crotonyl-CoA Carboxylase Deficiency Sanger Sequencing	81406
MG6054	Del-Dup Exonic Microarray	81479
MG54C	1 or 2 Known Mutation	81479
MG54P	Prenatal Sequencing incl. MCC	81406 & 81265 or 81266*
MG54P1	Prenatal 1 Known Mutation incl. MCC	81479 & 81265 or 81266*
MG54P2	Prenatal 2 Known Mutation incl. MCC	81479 & 81265 or 81266*
MG72	MCEE - Epimerase Deficiency Sanger Sequencing	81479
MG6072	Del-Dup Exonic Microarray	81479
MG72C	1 or 2 Known Mutation	81479
MG72P	Prenatal Sequencing incl. MCC	81479 & 81265 or 81266*
MG72P1	Prenatal 1 Known Mutation incl. MCC	81479 & 81265 or 81266*
MG72P2	Prenatal 2 Known Mutation incl. MCC	81479 & 81265 or 81266*
L7335	MECP2 - Rett Syndrome Sanger Sequencing	81302
L7336	Del-Dup Exonic Microarray	81304
MG285C	1 or 2 Known Mutation	81303
MG285P	Prenatal Sequencing incl. MCC	81302 & 81265 or 81266*
MG285P1	Prenatal 1 Known Mutation incl. MCC	81303 & 81265 or 81266*
MG285P2	Prenatal 2 Known Mutation incl. MCC	81303 & 81265 or 81266*
MG286	MEN1 - Multiple Endocrine Neoplasia Type 1 Sanger Sequencing	81405
MG6286	Del-Dup Exonic Microarray	81404
MG286C	1 or 2 Known Mutation	81479
MG286P	Prenatal Sequencing incl. MCC	81405 & 81265 or 81266*
MG286P1	Prenatal 1 Known Mutation incl. MCC	81479 & 81265 or 81266*
MG286P2	Prenatal 2 Known Mutation incl. MCC	81479 & 81265 or 81266*
MG34	MITF - Waardenburg Syndrome Sanger Sequencing	81479
MG6034	Del-Dup Exonic Microarray	81479
MG34C	1 or 2 Known Mutation	81479
MG34P	Prenatal Sequencing incl. MCC	81479 & 81265 or 81266*
MG34P1	Prenatal 1 Known Mutation incl. MCC	81479 & 81265 or 81266*
MG34P2	Prenatal 2 Known Mutation incl. MCC	81479 & 81265 or 81266*
MG68	MMAA - Methylmalonic Acidemia Sanger Sequencing	81405
MG6068	Del-Dup Exonic Microarray	81479
MG68C	1 or 2 Known Mutation	81479
MG68P	Prenatal Sequencing incl. MCC	81405 & 81265 or 81266*
MG68P1	Prenatal 1 Known Mutation incl. MCC	81479 & 81265 or 81266*
MG68P2	Prenatal 2 Known Mutation incl. MCC	81479 & 81265 or 81266*
MG69	MMAB - Methylmalonic Acidemia Sanger Sequencing	81405
MG6069	Del-Dup Exonic Microarray	81479
MG69C	1 or 2 Known Mutation	81479
MG69P	Prenatal Sequencing incl. MCC	81405 & 81265 or 81266*
MG69P1	Prenatal 1 Known Mutation incl. MCC	81479 & 81265 or 81266*
MG69P2	Prenatal 2 Known Mutation incl. MCC	81479 & 81265 or 81266*

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MG71	MMACHC - Cobalamin C Deficiency Sanger Sequencing	81404
MG6071	Del-Dup Exonic Microarray	81479
MG71C	1 or 2 Known Mutation	81479
MG71P	Prenatal Sequencing incl. MCC	81404 & 81265 or 81266*
MG71P1	Prenatal 1 Known Mutation incl. MCC	81479 & 81265 or 81266*
MG71P2	Prenatal 2 Known Mutation incl. MCC	81479 & 81265 or 81266*
MG251	MMADHC (C2orf25) - Cobalamin D Deficiency Sanger Sequencing	81479
MG6251	Del-Dup Exonic Microarray	81479
MG251C	1 or 2 Known Mutation	81479
MG251P	Prenatal Sequencing incl. MCC	81479 & 81265 or 81266*
MG251P1	Prenatal 1 Known Mutation incl. MCC	81479 & 81265 or 81266*
MG251P2	Prenatal 2 Known Mutation incl. MCC	81479 & 81265 or 81266*
MG67	MUT - Mutase Deficiency Sanger Sequencing	81406
MG6067	Del-Dup Exonic Microarray	81479
MG67C	1 or 2 Known Mutation	81479
MG67P	Prenatal Sequencing incl. MCC	81406 & 81265 or 81266*
MG67P1	Prenatal 1 Known Mutation incl. MCC	81479 & 81265 or 81266*
MG67P2	Prenatal 2 Known Mutation incl. MCC	81479 & 81265 or 81266*
MG153	NCF2 - Chronic Granulomatous Disease Sanger Sequencing	81479
MG6153	Del-Dup Exonic Microarray	81479
MG153C	1 or 2 Known Mutation	81479
MG153P	Prenatal Sequencing incl. MCC	81479 & 81265 or 81266*
MG153P1	Prenatal 1 Known Mutation incl. MCC	81479 & 81265 or 81266*
MG153P2	Prenatal 2 Known Mutation incl. MCC	81479 & 81265 or 81266*
MG154	NCF4 - Chronic Granulomatous Disease Sanger Sequencing - p40-PHOX	81479
MG6154	Del-Dup Exonic Microarray	81479
MG154C	1 or 2 Known Mutation	81479
MG154P	Prenatal Sequencing incl. MCC	81479 & 81265 or 81266*
MG154P1	Prenatal 1 Known Mutation incl. MCC	81479 & 81265 or 81266*
MG154P2	Prenatal 2 Known Mutation incl. MCC	81479 & 81265 or 81266*
MG244	NFU1 - Multiple Mitochondrial Dysfunctions Syndrome, MMDS Sanger Sequencing	81479
MG6244	Del-Dup Exonic Microarray	81479
MG244C	1 or 2 Known Mutation	81479
MG244P	Prenatal Sequencing incl. MCC	81479 & 81265 or 81266*
MG244P1	Prenatal 1 Known Mutation incl. MCC	81479 & 81265 or 81266*
MG244P2	Prenatal 2 Known Mutation incl. MCC	81479 & 81265 or 81266*
MG255	OTX2 - Microphthalmia, Pituitary Hormone Deficiency, Retinal Dystrophy Sanger Sequencing	81479
MG6255	Del-Dup Exonic Microarray	81479
MG255C	1 or 2 Known Mutation	81479
MG255P	Prenatal Sequencing incl. MCC	81479 & 81265 or 81266*
MG255P1	Prenatal 1 Known Mutation incl. MCC	81479 & 81265 or 81266*
MG255P2	Prenatal 2 Known Mutation incl. MCC	81479 & 81265 or 81266*
MG88	P-gene - Albinism/Oculocutaneous Albinism: OCA2 Sanger Sequencing	81479
MG6088	Del-Dup Exonic Microarray	81479
MG88C	1 or 2 Known Mutation	81479
MG88P	Prenatal Sequencing incl. MCC	81479 & 81265 or 81266*
MG88P1	Prenatal 1 Known Mutation incl. MCC	81479 & 81265 or 81266*
MG88P2	Prenatal 2 Known Mutation incl. MCC	81479 & 81265 or 81266*
MG147	Sanger Sequencing plus 2.7kb Deletion	81479
MG89	2.7kb Deletion Only	81479
MG89P	Prenatal 2.7kb Deletion Only, incl. MCC	81479 & 81265 or 81266*
MG33	PAX3 - Waardenburg Syndrome Sanger Sequencing	81479
MG6033	Del-Dup Exonic Microarray	81479
MG33C	1 or 2 Known Mutation	81479
MG33P	Prenatal Sequencing incl. MCC	81479 & 81265 or 81266*
MG33P1	Prenatal 1 Known Mutation incl. MCC	81479 & 81265 or 81266*
MG33P2	Prenatal 2 Known Mutation incl. MCC	81479 & 81265 or 81266*

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L6948 L6722 MG148C MG148P MG148P1 MG148P2	PAX6 - PAX6 Related Disorders: Isolated Aniridia, Anophthalmia, Gillespie Sanger Sequencing Del-Dup Exonic Microarray 1 or 2 Known Mutation Prenatal Sequencing incl. MCC Prenatal 1 Known Mutation incl. MCC Prenatal 2 Known Mutation incl. MCC	81479 81479 81479 81479 & 81265 or 81266* 81479 & 81265 or 81266* 81479 & 81265 or 81266*
L7462 MG6078 MG78C MG78P MG78P1 MG78P2	PCCA - Propionic Acidemia PCC Sanger Sequencing Del-Dup Exonic Microarray 1 or 2 Known Mutation Prenatal Sequencing incl. MCC Prenatal 1 Known Mutation incl. MCC Prenatal 2 Known Mutation incl. MCC	81406 81405 81479 81406 & 81265 or 81266* 81479 & 81265 or 81266* 81479 & 81265 or 81266*
L7463 MG6079 MG79C MG79P MG79P1 MG79P2	PCCB - Propionic Acidemia PCC Sanger Sequencing Del-Dup Exonic Microarray 1 or 2 Known Mutation Prenatal Sequencing incl. MCC Prenatal 1 Known Mutation incl. MCC Prenatal 2 Known Mutation incl. MCC	81406 81405 81479 81406 & 81265 or 81266* 81479 & 81265 or 81266* 81479 & 81265 or 81266*
MG127 MG6127 MG127C MG127P MG127P1 MG127P2	PDE6C - Achromatopsia Sanger Sequencing Del-Dup Exonic Microarray 1 or 2 Known Mutation Prenatal Sequencing incl. MCC Prenatal 1 Known Mutation incl. MCC Prenatal 2 Known Mutation incl. MCC	81479 81479 81479 81479 & 81265 or 81266* 81479 & 81265 or 81266* 81479 & 81265 or 81266*
L7426 MG6245 MG245C MG245P MG245P1 MG245P2	PDHA1 - Pyruvate Dehydrogenase Complex, PDH Sanger Sequencing Del-Dup Exonic Microarray 1 or 2 Known Mutation Prenatal Sequencing incl. MCC Prenatal 1 Known Mutation incl. MCC Prenatal 2 Known Mutation incl. MCC	81406 81405 81479 81406 & 81265 or 81266* 81479 & 81265 or 81266* 81479 & 81265 or 81266*
MG17 MG6017 MG17C MG17P MG17P1 MG17P2	PKP2 - Arrhythmogenic Right Ventricular Dysplasia (ARVD9) Sanger Sequencing Del-Dup Exonic Microarray 1 or 2 Known Mutation Prenatal Sequencing incl. MCC Prenatal 1 Known Mutation incl. MCC Prenatal 2 Known Mutation incl. MCC	81406 81479 81479 81406 & 81265 or 81266* 81479 & 81265 or 81266* 81479 & 81265 or 81266*
MG309 MG6039 MG309C MG309P MG309P1 MG309P2	PLCB4 - Auricularcondylar Syndrome 2 Sanger Sequencing Del-Dup Exonic Microarray 1 or 2 Known Mutation Prenatal Sequencing incl. MCC Prenatal 1 Known Mutation incl. MCC Prenatal 2 Known Mutation incl. MCC	81479 81479 81479 81479 & 81265 or 81266* 81479 & 81265 or 81266* 81479 & 81265 or 81266*
L7302 MG6155 MG155C MG155P MG155P1 MG155P2	PNPO - Pyridoxal Phosphate Dependent Neonatal Seizures Sanger Sequencing Del-Dup Exonic Microarray 1 or 2 Known Mutation Prenatal Sequencing incl. MCC Prenatal 1 Known Mutation incl. MCC Prenatal 2 Known Mutation incl. MCC	81479 81479 81479 81479 & 81265 or 81266* 81479 & 81265 or 81266* 81479 & 81265 or 81266*
L6911 L7280 L7281 MG103P MG103P1 MG103P2	POLG1 - POLG1 Related Disorders for Mitochondrial Disorders Sanger Sequencing Del-Dup Exonic Microarray 1 or 2 Known Mutation Prenatal Sequencing incl. MCC Prenatal 1 Known Mutation incl. MCC Prenatal 2 Known Mutation incl. MCC	81406 81479 81479 81406 & 81265 or 81266* 81479 & 81265 or 81266* 81479 & 81265 or 81266*
L6942 L6904 L6910 MG110P MG110P1 MG110P2	PTEN - PTEN Related Disorders Sanger Sequencing Del-Dup Exonic Microarray 1 or 2 Known Mutation Prenatal Sequencing incl. MCC Prenatal 1 Known Mutation incl. MCC Prenatal 2 Known Mutation incl. MCC	81321 81323 81322 81321 & 81265 or 81266* 81322 & 81265 or 81266* 81323 & 81265 or 81266*

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MG289	RB1 - Retinoblastoma Sanger Sequencing	81479
MG6289	Del-Dup Exonic Microarray	81479
MG289C	1 or 2 Known Mutation	81479
MG289P	Prenatal Sequencing incl. MCC	81479 & 81265 or 81266*
MG289P1	Prenatal 1 Known Mutation incl. MCC	81479 & 81265 or 81266*
MG289P2	Prenatal 2 Known Mutation incl. MCC	81479 & 81265 or 81266*
MG290	RET - Multiple Endocrine Neoplasia, Type 2A & 2B Sanger Sequencing	81406
MG6290	Del-Dup Exonic Microarray	81479
MG290C	1 or 2 Known Mutation	81479
MG290P	Prenatal Sequencing incl. MCC	81406 & 81265 or 81266*
MG290P1	Prenatal 1 Known Mutation incl. MCC	81479 & 81265 or 81266*
MG290P2	Prenatal 2 Known Mutation incl. MCC	81479 & 81265 or 81266*
MG99	RNASEH2A - Aicardi-Goutieres Syndrome (AGS) Sanger Sequencing	81479
MG6099	Del-Dup Exonic Microarray	81479
MG99C	1 or 2 Known Mutation	81479
MG99P	Prenatal Sequencing incl. MCC	81479 & 81265 or 81266*
MG99P1	Prenatal 1 Known Mutation incl. MCC	81479 & 81265 or 81266*
MG99P2	Prenatal 2 Known Mutation incl. MCC	81479 & 81265 or 81266*
MG97	RNASEH2B - Aicardi-Goutieres Syndrome (AGS) Sanger Sequencing	81479
MG6097	Del-Dup Exonic Microarray	81479
MG67C	1 or 2 Known Mutation	81479
MG97P	Prenatal Sequencing incl. MCC	81479 & 81265 or 81266*
MG97P1	Prenatal 1 Known Mutation incl. MCC	81479 & 81265 or 81266*
MG97P2	Prenatal 2 Known Mutation incl. MCC	81479 & 81265 or 81266*
MG98	RNASEH2C - Aicardi-Goutieres Syndrome (AGS) Sanger Sequencing	81479
MG6098	Del-Dup Exonic Microarray	81479
MG98C	1 or 2 Known Mutation	81479
MG98P	Prenatal Sequencing incl. MCC	81479 & 81265 or 81266*
MG98P1	Prenatal 1 Known Mutation incl. MCC	81479 & 81265 or 81266*
MG98P2	Prenatal 2 Known Mutation incl. MCC	81479 & 81265 or 81266*
MG252	RNASET2 - Cystic Leukoencephalopathy Sanger Sequencing	81479
MG6252	Del-Dup Exonic Microarray	81479
MG252C	1 or 2 Known Mutation	81479
MG252P	Prenatal Sequencing incl. MCC	81479 & 81265 or 81266*
MG252P1	Prenatal 1 Known Mutation incl. MCC	81479 & 81265 or 81266*
MG252P2	Prenatal 2 Known Mutation incl. MCC	81479 & 81265 or 81266*
L6920	SALL1 - Townes-Brock Syndrome Sanger Sequencing	81479
L7393	Del-Dup Exonic Microarray	81479
MG48C	1 or 2 Known Mutation	81479
MG48P	Prenatal Sequencing incl. MCC	81479 & 81265 or 81266*
MG48P1	Prenatal 1 Known Mutation incl. MCC	81479 & 81265 or 81266*
MG48P2	Prenatal 2 Known Mutation incl. MCC	81479 & 81265 or 81266*
L6932	SALL4 - Duane Radial Ray Syndrome Sanger Sequencing	81479
L7437	Del-Dup Exonic Microarray	81479
MG49C	1 or 2 Known Mutation	81479
MG49P	Prenatal Sequencing incl. MCC	81479 & 81265 or 81266*
MG49P1	Prenatal 1 Known Mutation incl. MCC	81479 & 81265 or 81266*
MG49P2	Prenatal 2 Known Mutation incl. MCC	81479 & 81265 or 81266*
MG123	SAMHD1 - Aicardi-Goutieres Syndrome (AGS) Sanger Sequencing	81479
MG6123	Del-Dup Exonic Microarray	81479
MG123C	1 or 2 Known Mutation	81479
MG123P	Prenatal Sequencing incl. MCC	81479 & 81265 or 81266*
MG123P1	Prenatal 1 Known Mutation incl. MCC	81479 & 81265 or 81266*
MG123P2	Prenatal 2 Known Mutation incl. MCC	81479 & 81265 or 81266*
MG308	SLC19A3 - Biotin-Responsive Basal Ganglia Disease Sanger Sequencing	81479
MG6308	Del-Dup Exonic Microarray	81479
MG308C	1 or 2 Known Mutation	81479
MG308P	Prenatal Sequencing incl. MCC	81479 & 81265 or 81266*
MG308P1	Prenatal 1 Known Mutation incl. MCC	81479 & 81265 or 81266*
MG308P2	Prenatal 2 Known Mutation incl. MCC	81479 & 81265 or 81266*

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L7355 L7356 MG256C MG256P MG256P1 MG256P2	SLC22A5 - Systemic Primary Carnitine Deficiency Sanger Sequencing Del-Dup Exonic Microarray 1 or 2 Known Mutation Prenatal Sequencing incl. MCC Prenatal 1 Known Mutation incl. MCC Prenatal 2 Known Mutation incl. MCC	81405 81479 81479 81405 & 81265 or 81266* 81479 & 81265 or 81266* 81479 & 81265 or 81266*
MG31 MG6031 MG31C MG31P MG31P1 MG31P2	SLC26A4 - Pendred Syndrome Sanger Sequencing Del-Dup Exonic Microarray 1 or 2 Known Mutation Prenatal Sequencing incl. MCC Prenatal 1 Known Mutation incl. MCC Prenatal 2 Known Mutation incl. MCC	81406 81479 81479 81406 & 81265 or 81266* 81479 & 81265 or 81266* 81479 & 81265 or 81266*
MG91 MG6091 MG91C MG91P MG91P1 MG91P2	SLC45A2 - Albinism/Oculocutaneous Albinism: OCA4 Sanger Sequencing Del-Dup Exonic Microarray 1 or 2 Known Mutation Prenatal Sequencing incl. MCC Prenatal 1 Known Mutation incl. MCC Prenatal 2 Known Mutation incl. MCC	81479 81479 81479 81479 & 81265 or 81266* 81479 & 81265 or 81266* 81479 & 81265 or 81266*
MG291 MG6291 MG291C MG291P MG291P1 MG291P2	SMAD4 - Hereditary Hemorrhagic Telangiectasia/Juvenile Polyposis Syndrome Sanger Sequencing Del-Dup Exonic Microarray 1 or 2 Known Mutation Prenatal Sequencing incl. MCC Prenatal 1 Known Mutation incl. MCC Prenatal 2 Known Mutation incl. MCC	81406 81405 81479 81406 & 81265 or 81266* 81479 & 81265 or 81266* 81479 & 81265 or 81266*
MG258 MG6258 MG258C MG258P MG258P1 MG258P2	SOX2 - Microphthalmia, Optic Nerve Hypoplasia Sanger Sequencing Del-Dup Exonic Microarray 1 or 2 Known Mutation Prenatal Sequencing incl. MCC Prenatal 1 Known Mutation incl. MCC Prenatal 2 Known Mutation incl. MCC	81479 81479 81479 81479 & 81265 or 81266* 81479 & 81265 or 81266* 81479 & 81265 or 81266*
L7450 MG6257 MG257C MG257P MG257P1 MG257P2	SOX9 - Campomelic Dysplasia with Autosomal Sex Reversal Sanger Sequencing Del-Dup Exonic Microarray 1 or 2 Known Mutation Prenatal Sequencing incl. MCC Prenatal 1 Known Mutation incl. MCC Prenatal 2 Known Mutation incl. MCC	81479 81479 81479 81479 & 81265 or 81266* 81479 & 81265 or 81266* 81479 & 81265 or 81266*
MG35 MG6035 MG35C MG35P MG35P1 MG35P2	SOX10 - Waardenburg Syndrome Sanger Sequencing Del-Dup Exonic Microarray 1 or 2 Known Mutation Prenatal Sequencing incl. MCC Prenatal 1 Known Mutation incl. MCC Prenatal 2 Known Mutation incl. MCC	81479 81479 81479 81479 & 81265 or 81266* 81479 & 81265 or 81266* 81479 & 81265 or 81266*
L7349 L7350 MG80C MG80P MG80P1 MG80P2	SPR - Sepiapterin Reductase Deficiency, SPR Sanger Sequencing Del-Dup Exonic Microarray 1 or 2 Known Mutation Prenatal Sequencing incl. MCC Prenatal 1 Known Mutation incl. MCC Prenatal 2 Known Mutation incl. MCC	81479 81479 81479 81479 & 81265 or 81266* 81479 & 81265 or 81266* 81479 & 81265 or 81266*
MG261 MG6261 MG261C MG261P MG261P1 MG261P2	SPRED1 - Neurofibromatosis-Like/Legius Syndrome Sanger Sequencing Del-Dup Exonic Microarray 1 or 2 Known Mutation Prenatal Sequencing incl. MCC Prenatal 1 Known Mutation incl. MCC Prenatal 2 Known Mutation incl. MCC	81405 81479 81479 81405 & 81265 or 81266* 81479 & 81265 or 81266* 81479 & 81265 or 81266*
L6916 L7292 L7362 MG39P MG39P1 MG39P2	SRY - 46, XY Disorder of Sexual Development Sanger Sequencing Del-Dup Exonic Microarray 1 or 2 Known Mutation Prenatal Sequencing incl. MCC Prenatal 1 Known Mutation incl. MCC Prenatal 2 Known Mutation incl. MCC	81400 81479 81479 81400 & 81265 or 81266* 81479 & 81265 or 81266* 81479 & 81265 or 81266*

*81266 will be applied for twin specimens

For custom Del-Dup or qPCR analysis please contact us

L6931 L7080 MG294C MG294P MG294P1 MG294P2	STK11 - Peutz-Jeghers Syndrome Sanger Sequencing Del-Dup Exonic Microarray 1 or 2 Known Mutation Prenatal Sequencing incl. MCC Prenatal 1 Known Mutation incl. MCC Prenatal 2 Known Mutation incl. MCC	81405 81404 81479 81405 & 81265 or 81266* 81479 & 81265 or 81266* 81479 & 81265 or 81266*
L7049 L7418 MG50C MG50P MG50P1 MG50P2	TBX5 - Holt-Oram Syndrome Sanger Sequencing Del-Dup Exonic Microarray 1 or 2 Known Mutation Prenatal Sequencing incl. MCC Prenatal 1 Known Mutation incl. MCC Prenatal 2 Known Mutation incl. MCC	81405 81404 81479 81405 & 81265 or 81266* 81479 & 81265 or 81266* 81479 & 81265 or 81266*
MG307 MG6307 MG307C MG307P MG307P1 MG307P2	TFAP2A - Branchioculofacial Syndrome Sanger Sequencing Del-Dup Exonic Microarray 1 or 2 Known Mutation Prenatal Sequencing incl. MCC Prenatal 1 Known Mutation incl. MCC Prenatal 2 Known Mutation incl. MCC	81479 81479 81479 81479 & 81265 or 81266* 81479 & 81265 or 81266* 81479 & 81265 or 81266*
L7043 MG6096 L7301 MG96P MG96P1 MG96P2	TREX1 - Aicardi-Goutieres Syndrome (AGS) Sanger Sequencing Del-Dup Exonic Microarray 1 or 2 Known Mutation Prenatal Sequencing incl. MCC Prenatal 1 Known Mutation incl. MCC Prenatal 2 Known Mutation incl. MCC	81479 81479 81479 81479 & 81265 or 81266* 81479 & 81265 or 81266* 81479 & 81265 or 81266*
L7040 L7041 MG259C MG259P MG259P1 MG259P2	TWIST1 - Saethre-Chotzen Syndrome Sanger Sequencing Del-Dup Exonic Microarray 1 or 2 Known Mutation Prenatal Sequencing incl. MCC Prenatal 1 Known Mutation incl. MCC Prenatal 2 Known Mutation incl. MCC	81404 81403 81479 81404 & 81265 or 81266* 81479 & 81265 or 81266* 81479 & 81265 or 81266*
MG87 MG6087 MG87C MG87P MG87P1 MG87P2	TYR - Albinism/Oculocutaneous Albinism: OCA1 Sanger Sequencing Del-Dup Exonic Microarray 1 or 2 Known Mutation Prenatal Sequencing incl. MCC Prenatal 1 Known Mutation incl. MCC Prenatal 2 Known Mutation incl. MCC	81404 81403 81479 81404 & 81265 or 81266* 81479 & 81265 or 81266* 81479 & 81265 or 81266*
MG90 MG6090 MG90C MG90P MG90P1 MG90P2	TYRP1 - Albinism/Oculocutaneous Albinism: OCA3 Sanger Sequencing, OCA3 Del-Dup Exonic Microarray 1 or 2 Known Mutation Prenatal Sequencing incl. MCC Prenatal 1 Known Mutation incl. MCC Prenatal 2 Known Mutation incl. MCC	81479 81479 81479 81479 & 81265 or 81266* 81479 & 81265 or 81266* 81479 & 81265 or 81266*
L7342 L7343 MG297C MG297P MG297P1 MG297P2	VHL - Von Hippel-Lindau Sanger Sequencing Del-Dup Exonic Microarray 1 or 2 Known Mutation Prenatal Sequencing incl. MCC Prenatal 1 Known Mutation incl. MCC Prenatal 2 Known Mutation incl. MCC	81404 81403 81479 81404 & 81265 or 81266* 81479 & 81265 or 81266* 81479 & 81265 or 81266*
MG137 MG6137 MG137C MG137P MG137P1 MG137P2	WFS1 - Wolfram Syndrome Sanger Sequencing, OCA3 Del-Dup Exonic Microarray 1 or 2 Known Mutation Prenatal Sequencing incl. MCC Prenatal 1 Known Mutation incl. MCC Prenatal 2 Known Mutation incl. MCC	81479 81479 81479 81479 & 81265 or 81266* 81479 & 81265 or 81266* 81479 & 81265 or 81266*
L6727 L7275 L7261 L6728 MG40P1 MG40P2	WT1 - 46, XY Disorder of Sexual Development/Fraser Syndrome/Denys-Drash Syndrome Sanger Sequencing Del-Dup Exonic Microarray 1 or 2 Known Mutation Prenatal Sequencing incl. MCC Prenatal 1 Known Mutation incl. MCC Prenatal 2 Known Mutation incl. MCC	81405 81479 81479 81405 & 81265 or 81266* 81479 & 81265 or 81266* 81479 & 81265 or 81266*

*81266 will be applied for twin specimens

For custom Del-Dup or qPCR analysis please contact us