

Multi-Divisional Tests

Test Code	Test Name	Buccal*	Oral Rinse	DBS
905	Del/Dup Familial	Yes	Yes	No
906	Del/Dup for 1 Gene	Yes	Yes	No
J774	GenomeSeqDx	No	No	No
9011	One Known Mutation	Yes	Yes	Yes
205	PTCH1 Gene Sequencing & Del/Dup	Yes	Yes	No
195	PTEN Gene Sequencing and Del/Dup	Yes	Yes	No
J842	ReproXpanded Couple	Yes	Yes	Yes
J776	ReproXpanded Individual	Yes	Yes	Yes
363	TTR Gene Sequencing	Yes	Yes	Yes
9012	Two Known Mutations	Yes	Yes	Yes
561	Exome Sequencing (XomeDx)	Yes	Yes	Yes
910	Whole Genome Array (GenomeDx)	Yes	Yes	No
J775	XomeDxInsights	No	No	No
690	XomeDxPlus (WES + mtDNA Sequencing and Deletion Testing)	Yes	Yes	No
706	XomeDxSlice (Phenotype-driven Specific Gene List Using WES Capture)	Yes	Yes	Yes
896 - Trio	XomeDxXpress (WES with a Verbal Result in 7 Days)	No	No	No

Oncology Tests

Test Code	Test Name	Buccal*	Oral Rinse	DBS
717	BMPR1A, SMAD4 Gene Sequencing & Del/Dup	Yes	Yes	No
B501	BRCA1 and BRCA2 Del/Dup	Yes	Yes	No
B502	BRCA1 and BRCA2 Sequencing	Yes	Yes	Yes
B361	BRCA1/2 Ashkenazi Founder Mutation Panel	Yes	Yes	No
B362	BRCA1/2 Sequencing and Del/Dup analysis	Yes	Yes	No
B521	Breast Cancer High Risk Panel	Yes	Yes	No
J005	Breast Cancer High Risk Panel and PALB2	Yes	Yes	No
J055	Breast Cancer High/Moderate Risk Panel	Yes	Yes	No
J662	Breast Cancer Surgical Panel	Yes	Yes	No
B273	Breast/Ovarian Cancer Panel	Yes	Yes	No
721	CDC73 Gene Sequencing and Del/Dup	Yes	Yes	No
2021	CDKN2A (p16) & CDK4 (Exon2) Sequencing	Yes	Yes	Yes
B274	Colorectal Cancer Panel	Yes	Yes	No
B275	Comprehensive Cancer Panel	Yes	Yes	No
B344	Endometrial Cancer Panel	Yes	Yes	No
B399	Familial Cutaneous Malignant Melanoma	Yes	Yes	No
713	FH Gene Sequencing & Del/Dup	Yes	Yes	No
714	FLCN Gene Sequencing and Del/Dup	Yes	Yes	No
B751	High/Moderate Risk Panel	Yes	Yes	No
B522	Lynch/Colorectal High Risk Panel	Yes	Yes	No
719	MEN1 Gene Sequencing and Del/Dup	Yes	Yes	No
J006	MSH2 Exon 1-7 Inversion Analysis	Yes	Yes	No

*denotes GeneDx Sponge Swab

Oncology Tests

Test Code	Test Name	Buccal*	Oral Rinse	DBS
B749	OncoGeneDx Custom Panel	Yes	Yes	No
B343	Pancreatic Cancer Panel	Yes	Yes	No
J318	Pediatric Tumor Panel	Yes	Yes	No
B395	PGL/PCC (Paranglioma/Pheochromocytoma) Panel	Yes	Yes	No
715	PRKAR1A Gene Sequencing & Del/Dup	Yes	Yes	No
J665	Prostate Cancer Panel	Yes	Yes	No
B394	Renal Cancer Panel	Yes	Yes	No
1771	RET Gene Sequencing	Yes	Yes	No
2071	STK11 Gene Sequencing & Del/Dup	Yes	Yes	No
718	TP53 Gene Sequencing & Del/Dup	Yes	Yes	No
332	VHL Gene Sequencing & Del/Dup	Yes	Yes	No

Cardiology Tests

Test Code	Test Name	Buccal*	Oral Rinse	DBS
483	ARVC Sequencing and Del/Dup Panel	Yes	Yes	No
481	BrS Sequencing and Del/Dup Panel	Yes	Yes	No
935	Combined Cardiac Panel	Yes	Yes	No
695	Comprehensive Arrhythmia Panel	Yes	Yes	No
694	Comprehensive Cardiomyopathy Panel	Yes	Yes	No
482	CPVT Sequencing and Del/Dup Panel	Yes	Yes	No
695C	Custom Arrhythmia Panel	Yes	Yes	No
483C	Custom Arrhythmogenic Right Ventricular Cardiomyopathy (ARVC) Panel	Yes	Yes	No
481C	Custom Brugada Syndrome (BrS)Panel	Yes	Yes	No
J779	Custom Cardiology Panel	Yes	Yes	No
694C	Custom Cardiomyopathy Panel	Yes	Yes	No
482C	Custom Catecholaminergic Polymorphic Ventricular Tachycardia (CPVT) Panel	Yes	Yes	No
935C	Custom Combined Cardiac Panel	Yes	Yes	No
J554C	Custom Dilated Cardiomyopathy (DCM)/Left Ventricular Non-Compaction Cardiomyopathy (LVNC) Panel	Yes	Yes	No
J556C	Custom Familial Hypercholesterolemia (FH) Panel	Yes	Yes	No
J555C	Custom Heritable Disorders of Connective Tissue (HDCT) Panel	Yes	Yes	No
J553C	Custom Hypertrophic Cardiomyopathy (HCM) Panel	Yes	Yes	No
727C	Custom Long QT Syndrome (LQTS) Panel	Yes	Yes	No
883C	Custom Marfan Syndrome/Thoracic Aortic Aneurysm and Dissection (TAAD) and Related Disorders Panel	Yes	Yes	No
J551C	Custom Short QT Syndrome (SQTS) Panel	Yes	Yes	No
J552C	Custom Sudden Cardiac Arrest (SCA) Panel	Yes	Yes	No
J554	DCM/LVNC Sequencing and Del/Dup Panel	Yes	Yes	No
401	ELN Gene Sequencing	Yes	Yes	No
J556	Familial Hypercholesterolemia Panel	Yes	Yes	No
918	FBN1 Gene Sequencing & Del/Dup	Yes	Yes	No
J553	HCM Sequencing and Del/Dup Panel	Yes	Yes	No
697	Hereditary Hemorrhagic Telangiectasia Panel	Yes	Yes	No
J555	Heritable Disorders of Connective Tissue Panel	Yes	Yes	No
1004	JAG1 Gene Sequencing & Del/Dup	Yes	Yes	No
727	LQTS Sequencing & Del/Dup Panel	Yes	Yes	No

Cardiology Tests

Test Code	Test Name	Buccal*	Oral Rinse	DBS
883	Marfan/TAAD Sequencing & Del/Dup Panel	Yes	Yes	No
534	Noonan and RASopathies Panel	Yes	Yes	No
696	Pulmonary Arterial Hypertension Panel	Yes	Yes	No
J552	SCA Arrhythmia Sequencing and Del/Dup Panel	Yes	Yes	No
J551	SQTS Sequencing and Del/Dup Panel	Yes	Yes	No
2361	TBX5 Gene Sequencing	Yes	Yes	No

Neurology Tests

Test Code	Test Name	Buccal*	Oral Rinse	DBS
704	58 mtDNA Point Mutations plus Large Deletions Panel	Yes	Yes	No
219	AAAS Gene Sequencing	Yes	Yes	No
270	ACADVL Gene Sequencing	Yes	Yes	No
551	ACTA1 Gene Sequencing & NEB Mutation Analysis	Yes	Yes	Yes
547	AGS Panel	Yes	Yes	No
566	Angelman Syndrome Methylation Analysis	Yes	Yes	No
673	ANKRD11 Gene Sequencing	Yes	Yes	No
820	AR Repeat Analysis	Yes	No	No
J762	Ataxia Xpanded Panel	Yes	Yes	Yes
952	Autism/ID Xpanded Panel	Yes	Yes	Yes
885	Axonal CMT Panel	Yes	Yes	No
294	BTD Gene Sequencing	Yes	Yes	No
526	Cerebral Caverosus Malformations (CCM) Panel	Yes	Yes	No
542	Childhood-Onset Epilepsy Panel	Yes	Yes	No
J778	CMT Panel	Yes	Yes	No
819	CNBP Repeat Analysis	Yes	No	No
615	Combined Mito Genome Plus Mito Nuclear Gene Panel	Yes	Yes	No
691	Comprehensive Brain Malformations Panel	Yes	Yes	No
523	Comprehensive Epilepsy Panel	Yes	Yes	No
941	Comprehensive Hereditary Spastic Paraplegia Panel	Yes	Yes	No
573	Comprehensive Mitochondrial Nuclear Gene Panel	Yes	Yes	No
961	Comprehensive NF Panel	Yes	Yes	No
945	Congenital Myasthenia Syndromes Panel	Yes	Yes	No
892	Congenital Myopathies & Muscular Dystrophies Panel	Yes	Yes	No
884	Core CMT Panel	Yes	Yes	No
698	Cortical Brain Malformations Panel	Yes	Yes	No
334	CPT2 Gene Sequencing	Yes	Yes	Yes
2923	CREBBP Sequencing & Del/Dup	Yes	Yes	No
886	Demyelinating CMT Panel	Yes	Yes	No
2502	DHCR7 Gene Sequencing	Yes	Yes	No
787	DMD Del/Dup	Yes	Yes	No
786	DMD Gene Sequencing	Yes	Yes	Yes
818	DMPK Repeat Analysis	Yes	No	No
953	Epilepsy Del/Dup Panel	Yes	Yes	No
921	EpiXpanded Panel	Yes	Yes	Yes
664	Fatty Acid Oxidation Sequencing Panel	Yes	Yes	Yes

Neurology Tests

Test Code	Test Name	Buccal*	Oral Rinse	DBS
522	FMR1 CGG Repeat Analysis	Yes	No	No
507	GALC Gene Sequencing & Del/Dup	Yes	Yes	No
399	GCDH Gene Sequencing	Yes	Yes	No
527	GCH1 Gene Sequencing & Del/Dup	Yes	Yes	No
218	GFAP Gene Sequencing	Yes	Yes	Yes
2321	GLA Gene Sequencing	Yes	Yes	Yes
238	GNE (M712T) Mutation Analysis	Yes	Yes	Yes
415E	GPC3 Gene Sequencing & Del/Dup (Females)	Yes	Yes	No
415	GPC3 Gene Sequencing (Males)	Yes	Yes	Yes
532	HADHA/HADHB Gene Sequencing	Yes	Yes	No
737	Hereditary Neuropathy Panel	Yes	Yes	No
944	Hereditary Spastic Paraplegia Related Inborn Errors of Metabolism Panel	Yes	Yes	No
474	HESX1 Gene Sequencing	Yes	Yes	Yes
519	HEXA Gene Sequencing	Yes	Yes	No
2371	Holoprosencephaly Panel & Del/Dup	Yes	Yes	No
665	Hyperammonemia, Urea Cycle and Transporter Defects Sequencing Panel	Yes	Yes	Yes
342	IGHMBP2 Gene Sequencing	Yes	Yes	No
553	IKBK (NEMO) Gene Sequencing & Del/Dup	Yes	No	No
541	Infantile Epilepsy Panel	Yes	Yes	No
701	Joubert Syndrome and Related Disorders Panel	Yes	Yes	No
583	KMT2D Gene Sequencing	Yes	Yes	No
576	Lactic Acidosis/Pyruvate Metabolism Nuclear Gene Panel	Yes	Yes	No
J853	Leukodystrophy Xpanded Panel	Yes	Yes	Yes
890	Limb-Girdle Muscular Dystrophy Panel	Yes	Yes	No
946	Lissencephaly Panel	Yes	Yes	No
2432	MCOLN1 Gene Sequencing	Yes	Yes	No
549	MECP2 Gene Sequencing & Del/Dup	Yes	Yes	No
J544	Metabolic Myopathy	Yes	Yes	Yes
578	Methylglutaconic Aciduria Nuclear Gene Panel	Yes	Yes	No
667	Methylmalonic Acidemia, Cobalamin Metabolism and Related Disorders Sequencing Panel	Yes	Yes	Yes
689	Microcephaly Panel	Yes	Yes	No
J511	Microcephaly Xpanded Panel	Yes	Yes	Yes
554	Mito Genome Sequencing & Deletion Testing	Yes	Yes	No
575	Mitochondrial Encephalopathy/Leigh Syndrome Nuclear Gene Panel	Yes	Yes	No
274	MMACHC Gene Sequencing	Yes	Yes	No
444	mtDNA Del/Dup Analysis	Yes	No	No
893	Myofibrillar Myopathy Panel	Yes	Yes	No
962	Neurofibromatosis (NF1) Panel	Yes	Yes	No
963	Neurofibromatosis (NF2) Panel	Yes	Yes	No
889	Neuromuscular Disorders Panel	Yes	Yes	No
J660	NF1 Sequencing & Del/Dup	Yes	Yes	No
584	NIPBL and SMC1A Sequencing and Del/Dup Panel	Yes	Yes	No
581	NPC1/NPC2 Gene Sequencing	Yes	Yes	No
406	NSD1 Gene Sequencing & Del/Dup	Yes	Yes	No
239	NTRK1 Gene Sequencing	Yes	Yes	No
9017	One mtDNA mutation without heteroplasmy	Yes	Yes	No

Neurology Tests

Test Code	Test Name	Buccal*	Oral Rinse	DBS
453	One to Three mtDNA mutations with heteroplasmy	Yes	Yes	No
313E	OTC Gene Sequencing & Del/Dup (Females)	Yes	Yes	No
313	OTC Gene Sequencing (Males)	Yes	Yes	No
743	PABPN1 Repeat Analysis	Yes	Yes	No
742	PMP22 Del/Dup	Yes	Yes	No
888	PMP22 Gene Sequencing	Yes	Yes	No
394	POLG Gene Sequencing	Yes	Yes	No
700	Pontocerebellar Hypoplasia Panel	Yes	Yes	No
595	Prader-Willi Syndrome Methylation Analysis	Yes	Yes	No
577	Progressive External Ophthalmoplegia (PEO)/Optic Atrophy Nuclear Gene Panel	Yes	Yes	No
544	Progressive Myoclonic Epilepsy Panel	Yes	Yes	No
651	PRRT2 Gene Sequencing	Yes	Yes	No
557	PUS1 Gene Sequencing	Yes	Yes	No
2511	RAI1 Gene Sequencing & Del/Dup	Yes	Yes	No
683	Reflex Del/Dup Testing After Fatty Acid Oxidation Disorders Panel	Yes	Yes	No
684	Reflex Del/Dup Testing After Hyperammonemia Panel	Yes	Yes	No
J698	Reflex Del/Dup Testing After Metabolic Myopathy	Yes	Yes	No
685	Reflex Del/Dup Testing After MMA and Related Disorders Panel	Yes	Yes	No
729	Rett/Angelman Syndrome Panel	Yes	Yes	No
550	RSK2 (RPS6KA3) Gene Sequencing & Del/Dup	Yes	Yes	No
650	SCN9A Gene Sequencing	Yes	Yes	No
582	SDHA Gene Sequencing	Yes	Yes	No
375	SLC9A6 Gene Sequencing	Yes	Yes	No
2631	SMPD1 Gene Sequencing	Yes	Yes	No
814	STAT Epilepsy Panel	Yes	Yes	No
891	Syndromic Congenital Muscular Dystrophy Panel	Yes	Yes	No
699	Syndromic Macrocephaly/Overgrowth Panel	Yes	Yes	No
359	TH Gene Sequencing	Yes	Yes	No
607	TPP1 Gene Sequencing	Yes	Yes	No
730	Tuberous Sclerosis Panel	Yes	Yes	No
9020	Two mtDNA mutations without heteroplasmy	Yes	Yes	No
546	UBE3A Gene Sequencing & Del/Dup	Yes	Yes	No
942	Uncomplicated Hereditary Spastic Paraplegia Panel	Yes	Yes	No
227	VPS13B (COH1) Gene Sequencing	Yes	Yes	No

All Other Tests

Test Code	Test Name	Buccal*	Oral Rinse	DBS
120	ABCA12 Gene Sequencing	Yes	Yes	No
126	ABCA12 Select Exon Sequencing	Yes	Yes	No
2642	ABCC6 Gene Sequencing	Yes	Yes	No
2641	ABCC6 Hot Spots (Tier I)	Yes	Yes	No
114	ABHD5 Gene Sequencing	Yes	Yes	Yes
351	ACAD8 Gene Sequencing	Yes	Yes	Yes
2682	ACADM Gene Sequencing	Yes	Yes	Yes
456	ACADM Mutation Detection (T121I)	Yes	Yes	Yes

All Other Tests

Test Code	Test Name	Buccal*	Oral Rinse	DBS
2683	ACADM Remaining Exons Sequencing	Yes	Yes	Yes
2681	ACADM Select Exons Sequencing	Yes	Yes	Yes
269	ACADS Gene Sequencing	Yes	Yes	No
529	ACADSB (M389V) Mutation Analysis	Yes	Yes	No
383	ACADSB Gene Sequencing	Yes	Yes	No
354	ACAT1 Gene Sequencing	Yes	Yes	No
659	ACSF3 Gene Sequencing	Yes	Yes	No
244	ACTA1 Gene Sequencing	Yes	Yes	Yes
352	ADA Gene Sequencing	Yes	Yes	Yes
2971	adRP Tier 1	Yes	Yes	Yes
2975	adRP Tier 2	Yes	Yes	No
2974	adRP Tier 3	Yes	Yes	Yes
658	AGA Gene Sequencing	Yes	Yes	No
4701	AGS Tier 1	Yes	Yes	No
4702	AGS Tier 2	Yes	Yes	No
318	AICDA Gene Sequencing	Yes	Yes	No
379	AIPL1 Gene Sequencing	Yes	Yes	No
1393	AIRE Gene Sequencing	Yes	Yes	No
1392	AIRE Remaining Exons	Yes	Yes	No
1391	AIRE Select Exons	Yes	Yes	No
128	ALDH3A2 Gene Sequencing	Yes	Yes	No
1151	ALOX12B Gene Sequencing	Yes	Yes	No
1152	ALOXE3 Gene Sequencing	Yes	Yes	No
220	AR Gene Sequencing	Yes	Yes	Yes
465	ARG1 Gene Sequencing	Yes	Yes	No
908	arRP Del/Dup Panel	Yes	No	No
368	arRP Sequencing Panel	Yes	No	No
563	ARSA Gene Sequencing	Yes	Yes	No
565	ARSB Gene Sequencing	Yes	Yes	No
282	ARSE Gene Sequencing (Male)	Yes	Yes	Yes
282E	ARSE Gene Sequencing & Del/Dup (Females)	Yes	Yes	No
426	ASL Gene Sequencing	Yes	Yes	No
564	ASPA Gene Sequencing & Del/Dup	Yes	Yes	No
382	ASS1 Gene Sequencing	Yes	Yes	No
201	ATP2A2 Gene Sequencing	Yes	Yes	No
206	ATP2C1 Gene Sequencing	Yes	Yes	No
501	AUH Gene Sequencing	Yes	Yes	No
4881	BCKDHA Gene Sequencing	Yes	Yes	No
4882	BCKDHB Gene Sequencing	Yes	Yes	No
3693	BCOR Gene Sequencing & Del/Dup	Yes	Yes	No
370	BCOR P85L Mutation	Yes	Yes	Yes
3692	BCOR Remaining Exons Sequencing	Yes	Yes	Yes
3691	BCOR Select Exons Sequencing & Del/Dup	Yes	Yes	No
403	BEST1 Gene Sequencing	Yes	Yes	No
372	BLM Gene Sequencing	Yes	Yes	No
603	B-negative SCID Panel	Yes	Yes	No

All Other Tests

Test Code	Test Name	Buccal*	Oral Rinse	DBS
602	B-positive SCID Panel	Yes	Yes	No
154	BTK Gene Sequencing & Del/Dup	Yes	Yes	No
590	CABP4 Gene Sequencing	Yes	Yes	No
587	CACNA1F Gene Sequencing	Yes	Yes	No
2611	CASP10 Gene Sequencing	Yes	Yes	Yes
2612	CASP8 Gene Sequencing	Yes	Yes	No
170	CASR Gene Sequencing	Yes	Yes	Yes
535	CBL/NRAS Gene Sequencing	Yes	Yes	No
331	CBS Gene Sequencing	Yes	Yes	No
4181	CCM Tier 1	Yes	Yes	No
4182	CCM Tier 2	Yes	Yes	No
419	CCM2 Gene Sequencing	Yes	Yes	No
668	CD40 Gene Sequencing	Yes	Yes	No
669	CD40LG Gene Sequencing	Yes	Yes	No
173	CDC73 (HRPT2) Gene Sequencing	Yes	Yes	No
1732	CDC73 (HRPT2) Remaining Exons Sequencing	Yes	Yes	No
1731	CDC73 (HRPT2) Select Exons Sequencing	Yes	Yes	No
3051	CDKL5 Gene Sequencing	Yes	Yes	No
2022	CDKN2A (p16) Gene Sequencing	Yes	Yes	Yes
376	CEP290 Hot Spots	Yes	Yes	No
506	CERKL Gene Sequencing	Yes	Yes	No
1431	CGD Tier 1	Yes	Yes	No
2261	CHD7 Gene Sequencing	Yes	Yes	No
296	CHM Gene Sequencing	Yes	Yes	No
229	CLCN5 Gene Sequencing	Yes	Yes	Yes
417	CNGA1 Gene Sequencing	Yes	Yes	Yes
514	CNGA3 Gene Sequencing	Yes	Yes	Yes
513	CNGB3 Gene Sequencing	Yes	Yes	No
1636	COL17A1 Gene Sequencing	Yes	Yes	No
281	COL4A5 Gene Sequencing	Yes	Yes	No
162	COL7A1 Gene Sequencing	Yes	Yes	No
249	COMP Gene Sequencing	Yes	Yes	Yes
601	Comprehensive SCID Panel	Yes	Yes	No
468	Cone-Rod Dystrophy Sequencing Panel	Yes	No	No
708	Congenital Ichthyosis XomeDxSlice	Yes	Yes	Yes
938	Congenital Sideroblastic Anemia Panel	No	No	No
119	Connexin Gene Testing	Yes	Yes	Yes
425	CPT1A Gene Sequencing	Yes	Yes	No
378	CRB1 Gene Sequencing	Yes	Yes	No
2922	CREBBP Remaining Exons Sequencing	Yes	Yes	No
2921	CREBBP Select Exons Sequencing & Del/Dup	Yes	Yes	No
353	CRX Gene Sequencing	Yes	Yes	No
735	CTSC Gene Sequencing	Yes	Yes	Yes
703	Custom ExonArrayDx	Yes	Yes	No
1436	CYBA Gene Sequencing	Yes	Yes	No
1434	CYBB Gene Sequencing	Yes	Yes	No

All Other Tests

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402	CYP17A1 Gene Sequencing	Yes	Yes	No
340	CYP19A1 Gene Sequencing	Yes	Yes	No
330	CYP1B1 Gene Sequencing	Yes	Yes	Yes
185	CYP27B1 Select Exons Sequencing	Yes	Yes	Yes
637	CYP4F22 Gene Sequencing	Yes	Yes	Yes
4883	DBT Gene Sequencing	Yes	Yes	No
1502	DCLRE1C Athabaskan mutation	Yes	Yes	No
1501	DCLRE1C Gene Sequencing & Deletion	Yes	Yes	No
422	DHH Gene Sequencing	Yes	Yes	No
J450	Diamond Blackfan Anemia Panel	Yes	Yes	No
108	DKC1 Gene Sequencing	Yes	Yes	No
490	DLD Gene Sequencing	Yes	Yes	No
314	DMP1 Gene Sequencing	Yes	Yes	Yes
679	DOCK8 Gene Sequencing	Yes	Yes	No
736	DOCK8 Gene Sequencing and Del/Dup	Yes	No	No
1601E	EDA1 Gene Sequencing & Del/Dup (Female)	Yes	Yes	No
1601	EDA1 Gene Sequencing (Males)	Yes	Yes	Yes
156	EDAR Gene Sequencing	Yes	Yes	Yes
617	EDARADD Gene Sequencing	Yes	Yes	Yes
3251	EFNB1 Gene Sequencing	Yes	Yes	Yes
105	ELANE (ELA2) Gene Sequencing	Yes	Yes	Yes
2990	ELOVL4 Tier 1	Yes	Yes	Yes
2991	ELOVL4 Tier 2	Yes	Yes	Yes
707	Epidermolysis Bullosa (EB) XomeDxSlice	Yes	Yes	Yes
278	ETFA Gene Sequencing	Yes	Yes	No
279	ETFB Gene Sequencing	Yes	Yes	No
280	ETFDH Gene Sequencing	Yes	Yes	No
558	ETHE1 Gene Sequencing & Del/Dup	Yes	Yes	No
1813	EXT1 & EXT2 Sequencing & Del/Dup	Yes	Yes	No
315E	EYA1 Gene Sequencing & Del/Dup	Yes	Yes	No
388	F12 Select Exons Sequencing	Yes	Yes	Yes
3661	FAH Gene Sequencing	Yes	Yes	No
3663	FAH Remaining Exons Sequencing	Yes	Yes	No
3662	FAH Select Exons Sequencing	Yes	Yes	No
138	FAS (TNFRSF6) Gene Sequencing	Yes	Yes	No
184	FGF23 Gene Sequencing	Yes	Yes	Yes
2402	FGFR1 Gene Sequencing	Yes	Yes	No
2843	FH Gene Sequencing	Yes	Yes	No
2842	FH Remaining Exons Sequencing	Yes	Yes	No
2841	FH Select Exons Sequencing	Yes	Yes	No
336	FISH Analysis	No	No	No
197	FLCN Gene Sequencing	Yes	Yes	Yes
123	FLG (Filaggrin) Hot Spots	Yes	Yes	Yes
1342	FOXC1 Gene Sequencing	Yes	Yes	Yes
604	FOXO3 Gene Sequencing	Yes	Yes	No
432	FRMD7 Gene Sequencing	Yes	Yes	No

All Other Tests

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661	FUCA1 Gene Sequencing	Yes	Yes	No
3271	FZD4 Gene Sequencing	Yes	Yes	Yes
287	GAA Gene Sequencing	Yes	Yes	No
499	GALK1 Gene Sequencing	Yes	Yes	No
608	GALNS Gene Sequencing	Yes	Yes	No
349E	GALT Gene Sequencing & Del/Dup	Yes	Yes	No
230	GCH1 Gene Sequencing	Yes	Yes	No
130	GJB2 Gene Sequencing	Yes	Yes	Yes
157	GJB6 Gene Sequencing	Yes	Yes	Yes
438	GK Gene Sequencing	Yes	Yes	No
657	GLB1 Gene Sequencing	Yes	Yes	No
472	GLI3 Gene Sequencing & Del/Dup	Yes	Yes	No
4712	GLI3 Remaining Exons Sequencing & Del/Dup	Yes	Yes	No
4711	GLI3 Select Exons Sequencing	Yes	Yes	No
589	GNAT1 Gene Sequencing	Yes	Yes	No
609	GNS Gene Sequencing & Del/Dup	Yes	Yes	No
588	GRM6 Gene Sequencing	Yes	Yes	No
476	GUCA1A Gene Sequencing	Yes	Yes	Yes
377	GUCY2D Gene Sequencing	Yes	Yes	No
467	GUCY2D Select Exons Sequencing	Yes	Yes	No
508	HADH Gene Sequencing	Yes	Yes	No
2711	HADHA Common Mutation (c.1528 G>C)	Yes	Yes	Yes
2712	HADHA Gene Sequencing	Yes	Yes	No
272	HADHB Gene Sequencing	Yes	Yes	No
303	HAX1 Gene Sequencing	Yes	Yes	Yes
515	HEXB Gene Sequencing & Del/Dup	Yes	Yes	No
593	HGSNAT Gene Sequencing	Yes	Yes	No
320	HLCS Gene Sequencing	Yes	Yes	No
1811	HME Tier 1	Yes	Yes	No
1812	HME Tier 2	Yes	Yes	No
3212	HMGCL Exon 2 Sequencing	Yes	Yes	Yes
3211	HMGCL Gene Sequencing	Yes	Yes	No
3213	HMGCL Remaining Exons Sequencing	Yes	Yes	Yes
503	HOXD13 Gene Sequencing	Yes	Yes	No
495	HPD Gene Sequencing	Yes	Yes	Yes
188	HPS1 & HPS3 Mutation Analysis (Puerto Rican)	Yes	Yes	Yes
189	HPS3 Mutation Analysis (Ashkenazi Jewish)	Yes	Yes	Yes
191	HRAS Gene Sequencing	Yes	Yes	Yes
463	HSD17B10 Gene Sequencing	Yes	Yes	Yes
678	Hyper-IgE Syndromes Panel	Yes	Yes	No
676	Hypogonadotropic Hypogonadism Gene Sequencing and Del/Dup Panel	Yes	Yes	No
1153	Ichthyin (NIPAL4) Gene Sequencing	Yes	Yes	Yes
2861	IKBKG (NEMO) Common Deletion (Female)	Yes	No	No
2863	IKBKG (NEMO) Gene Sequencing (HED)	Yes	No	No
2862	IKBKG (NEMO) Gene Sequencing (IP)	Yes	No	No
492	IL2RG Gene Sequencing	Yes	Yes	Yes

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302	IL7R Gene Sequencing	Yes	Yes	Yes
412	IMPDH1 Gene Sequencing	Yes	Yes	No
301	IRAK4 Gene Sequencing	Yes	Yes	Yes
248	IRF6 Exon 4 Sequencing	Yes	Yes	Yes
253	IRF6 Gene Sequencing	Yes	Yes	Yes
1642	ITGA6 Gene Sequencing	Yes	Yes	No
146	ITGB2 Gene Sequencing	Yes	Yes	No
1641	ITGB4 Gene Sequencing	Yes	Yes	No
3191	IVD Gene Sequencing	Yes	Yes	No
3192	IVD Tier 1	Yes	Yes	Yes
3193	IVD Tier 2	Yes	Yes	Yes
1001	JAG1 Tier 1	Yes	Yes	No
1002	JAG1 Tier 2	Yes	Yes	No
145	JAK3 Gene Sequencing	Yes	Yes	No
538	JPS Del/Dup Analysis	Yes	Yes	No
536	JPS Tier 1	Yes	Yes	No
537	JPS Tier 2	Yes	Yes	No
2401	KAL1 Gene Sequencing	Yes	Yes	No
2122	KRT1 Gene Sequencing	Yes	Yes	No
1182	KRT1 Gene Sequencing (EI)	Yes	Yes	No
1181	KRT1, KRT10 Hot Spots	Yes	Yes	No
1183	KRT10 Gene Sequencing	Yes	Yes	No
2133	KRT13 Gene Sequencing	Yes	Yes	No
1682	KRT14 Gene Sequencing	Yes	Yes	No
2121	KRT16 Hot Spots	Yes	Yes	No
2111	KRT17 Hot Spots	Yes	Yes	No
122	KRT2 Hot Spots	Yes	Yes	No
2132	KRT4 Gene Sequencing	Yes	Yes	No
2131	KRT4, KRT13 Hot Spots	Yes	Yes	No
1681	KRT5 Gene Sequencing	Yes	Yes	No
168	KRT5/14 Hot Spots	Yes	Yes	No
2094	KRT6A Gene Sequencing	Yes	Yes	No
2091	KRT6A, KRT16 Hot Spots	Yes	Yes	No
2096	KRT6B Gene Sequencing	Yes	Yes	No
2092	KRT6B, KRT17 Hot Spots	Yes	Yes	No
208	KRT9 Hot Spots	Yes	Yes	No
2551	L1CAM Gene Sequencing (Males)	Yes	Yes	No
1631	LAM5 Hot Spots	Yes	Yes	No
1634	LAMA3 Gene Sequencing	Yes	Yes	No
1632	LAMB3 Gene Sequencing	Yes	Yes	No
1633	LAMC2 Gene Sequencing	Yes	Yes	No
2980	LCA Tier 1	Yes	Yes	No
2981	LCA Tier 2	Yes	Yes	No
2982	LCA Tier 3	Yes	Yes	No
2983	LCA Tier 4	Yes	Yes	No
2984	LCA Tier 5	Yes	Yes	No

All Other Tests

Test Code	Test Name	Buccal*	Oral Rinse	DBS
2985	LCA Tier 6	Yes	Yes	No
655	LIPA Gene Sequencing	Yes	Yes	No
511	Loeys Dietz Syndrome Panel	Yes	Yes	No
3272	LRP5 Gene Sequencing	Yes	Yes	No
473	MAT1A Gene Sequencing	Yes	Yes	No
693	MBTPS2 Gene Sequencing	Yes	Yes	No
2882	MCCC1 Gene Sequencing	Yes	Yes	No
2881	MCCC2 Gene Sequencing	Yes	Yes	No
214	MEFV Select Exons Sequencing	Yes	Yes	Yes
176	MEN1 Gene Sequencing	Yes	Yes	No
404	MLYCD Gene Sequencing	Yes	Yes	No
276	MMAA Gene Sequencing	Yes	Yes	No
277	MMAB Gene Sequencing	Yes	Yes	No
674	MODY Sequencing Panel	Yes	Yes	No
104	MPL Gene Sequencing	Yes	Yes	Yes
488	MSUD Sequencing panel	Yes	Yes	No
2753	MUT Exon 2 Sequencing	Yes	Yes	No
2752	MUT Gene Sequencing	Yes	Yes	No
2754	MUT Remaining Exons Sequencing	Yes	Yes	No
2161	MVK Remaining Exons	Yes	Yes	Yes
216	MVK Select Exons	Yes	Yes	Yes
260	MYCN Gene Sequencing	Yes	Yes	Yes
329	MYOC Gene Sequencing	Yes	Yes	Yes
592	NAGLU Gene Sequencing	Yes	Yes	No
478	NAGS Gene Sequencing	Yes	Yes	No
1435	NCF1 (Exon 2) Gene Sequencing	Yes	Yes	No
1437	NCF2 Gene Sequencing	Yes	Yes	No
3273	NDP Gene Sequencing	Yes	Yes	Yes
3274	NDP Gene Sequencing & Del/Dup (Females)	Yes	Yes	No
245	NEB Exon 55 Deletion	Yes	Yes	Yes
648	NEU1 Gene Sequencing	Yes	Yes	No
569	NIPBL Remaining Exons Sequencing	Yes	Yes	No
568	NIPBL Select Exons Sequencing	Yes	Yes	No
217	NLRP3 Exon 3 Sequencing	Yes	Yes	Yes
246	NPC1 Gene Sequencing	Yes	Yes	No
247	NPC2 Gene Sequencing	Yes	Yes	No
416	NR0B1 (DAX1) Gene Sequencing	Yes	Yes	Yes
586	NR2E3 Gene Sequencing	Yes	Yes	Yes
341	NR5A1 Gene Sequencing	Yes	Yes	Yes
431	NYX Gene Sequencing	Yes	Yes	Yes
335	OCRL Gene Sequencing	Yes	Yes	No
3642	OFD1 Remaining Exons Sequencing	Yes	Yes	No
3641	OFD1 Select Exons Sequencing	Yes	Yes	No
346	OPTN Gene Sequencing	Yes	Yes	No
343	OTX2 Gene Sequencing	Yes	Yes	Yes
273	PAH Gene Sequencing	Yes	Yes	No

All Other Tests

Test Code	Test Name	Buccal*	Oral Rinse	DBS
5213	PAX2 Gene Sequencing	Yes	Yes	No
5211	PAX2 Tier 1	Yes	Yes	No
5212	PAX2 Tier 2	Yes	Yes	No
491	PAX6 Gene Sequencing & Del/Dup Panel	Yes	Yes	No
540	PC Gene Sequencing	Yes	Yes	No
2902	PCCA Gene Sequencing	Yes	Yes	No
2901	PCCB Gene Sequencing	Yes	Yes	No
420	PDCD10 Gene Sequencing	Yes	Yes	No
616	PDE6B Gene Sequencing	Yes	Yes	No
461	PDHA1 Gene Sequencing	Yes	Yes	No
462	PDHB Gene Sequencing	Yes	Yes	No
761	PEPD Gene Sequencing	Yes	Yes	No
367	Periodic Fever Syndromes Panel (7 genes)	Yes	Yes	No
1861E	PHEX Gene Sequencing & Del/Dup (Females)	Yes	Yes	No
1861	PHEX Gene Sequencing (Males)	Yes	Yes	No
1341	PITX2 Gene Sequencing	Yes	Yes	Yes
3482	PLEC1 Remaining Exons Sequencing	Yes	Yes	No
3481	PLEC1 Select Exons Sequencing	Yes	Yes	No
339	POR Gene Sequencing	Yes	Yes	No
306	PORCN Gene Sequencing & Del/Dup	Yes	Yes	No
677	Premature Ovarian Failure Sequencing Panel	Yes	Yes	No
198	PRKAR1A Gene Sequencing	Yes	Yes	Yes
2973	PRPF3 Gene Sequencing	Yes	Yes	No
300	PRPF8 Select Exons Sequencing	Yes	Yes	No
528	PSAP Gene Sequencing	Yes	Yes	No
2102	PSTPIP1 Remaining Exons Sequencing	Yes	Yes	No
2101	PSTPIP1 Select Exons Sequencing	Yes	Yes	No
1954	PTEN Promoter Sequencing	Yes	Yes	No
1953	PTEN Promoter Sequencing and Del/Dup	Yes	Yes	No
192	PTPN11 Gene Sequencing	Yes	Yes	No
380	PTS Gene Sequencing	Yes	Yes	No
649	PYGM Gene Sequencing	Yes	Yes	No
381	QDPR Gene Sequencing	Yes	Yes	No
147	RAG1/RAG2 Gene Sequencing	Yes	Yes	No
509	RAX Gene Sequencing	Yes	Yes	No
427	RDH5 Gene Sequencing	Yes	Yes	No
299	RDS (PRPH2) Gene Sequencing	Yes	Yes	No
400	Rest of the Periodic fever panel	Yes	Yes	No
2352	RET Remaining Exons Sequencing (HSCR)	Yes	Yes	No
1793	RET Remaining Exons Sequencing (MEN2A & MEN2B)	Yes	Yes	No
1791	RET Remaining Exons Sequencing (MEN2A)	Yes	Yes	No
1792	RET Remaining Exons Sequencing (MEN2B)	Yes	Yes	No
2351	RET Select Exons Sequencing (HSCR)	Yes	Yes	No
177	RET Select Exons Sequencing (MEN2A)	Yes	Yes	No
178	RET Select Exons Sequencing (MEN2B)	Yes	Yes	No
298	RHO Gene Sequencing	Yes	Yes	No

All Other Tests

Test Code	Test Name	Buccal*	Oral Rinse	DBS
815	RIT1 Gene Sequencing	Yes	Yes	Yes
4242	RLBP1 (BRD) Hot Spots	Yes	Yes	Yes
4243	RLBP1 (NFRCD) Hot Spots	Yes	Yes	Yes
4241	RLBP1 Gene Sequencing	Yes	Yes	Yes
225	RMRP Gene Sequencing	Yes	Yes	Yes
4703	RNASEH2C Hot Spots (Pakistani)	Yes	Yes	Yes
295	RP1 Hot Spots	Yes	Yes	No
326	RP2 Gene Sequencing	Yes	Yes	No
345	RPE65 Gene Sequencing	Yes	Yes	No
362	RPL11 Gene Sequencing	Yes	Yes	No
361	RPL5 Gene Sequencing	Yes	Yes	No
1061	RPS19 Gene Sequencing	Yes	Yes	No
1104	RSK2 (RPS6KA3) Gene Sequencing	Yes	Yes	No
1102	RSK2 (RPS6KA3) Remaining Exons Sequencing	Yes	Yes	No
1101	RSK2 (RPS6KA3) Select Exons Sequencing	Yes	Yes	No
517	SAG c.926delA Mutation	Yes	Yes	No
518	SAG Remaining Exon Sequencing	Yes	Yes	No
2521	SALL1 Gene Sequencing	Yes	Yes	No
262E	SALL4 Gene Sequencing & Del/Dup	Yes	Yes	No
610	Sanfilippo/MPSIII Sequencing Panel	Yes	Yes	No
109	SBDS Gene Sequencing	Yes	Yes	Yes
454	SDHAF2 (G78R) Mutation Analysis	Yes	Yes	No
322	SDHB Gene Sequencing	Yes	Yes	No
323	SDHC Gene Sequencing	Yes	Yes	No
324	SDHD Gene Sequencing	Yes	Yes	No
2341	SERPING1 (C1INH) Gene Sequencing & Del/Dup	Yes	Yes	No
591	SGSH Gene Sequencing	Yes	Yes	No
285	SH3BP2 Gene Sequencing	Yes	Yes	No
389	SHOC2 Gene Sequencing	Yes	Yes	Yes
317	SIX1 Gene Sequencing	Yes	Yes	Yes
605	SLC17A5 Gene Sequencing & Del/Dup	Yes	Yes	No
365	SLC22A5 Gene Sequencing	Yes	Yes	No
500	SLC25A13 Gene Sequencing	Yes	Yes	No
429	SLC25A20 Gene Sequencing	Yes	Yes	No
572	SLC26A4 Gene Sequencing	Yes	Yes	No
712	SLC27A4 Gene Sequencing	Yes	Yes	No
570	SMC1A Gene Sequencing	Yes	Yes	No
2632	SMPD1 Hot Spots (Ashkenazi Jewish mutation)	Yes	Yes	No
132	SOX2 Gene Sequencing	Yes	Yes	Yes
338	SOX9 Gene Sequencing	Yes	Yes	Yes
127	SPINK5 Gene Sequencing	Yes	Yes	No
816	SPRED1 Gene Sequencing	Yes	Yes	Yes
469	SRD5A2 Gene Sequencing	Yes	Yes	Yes
259	SRY Gene Sequencing	Yes	Yes	Yes
908	Stargardt Del/Dup Panel	Yes	Yes	No
466	Stargardt Sequencing Panel	Yes	Yes	No

All Other Tests

Test Code	Test Name	Buccal*	Oral Rinse	DBS
3123	STAT3 Gene Sequencing	Yes	Yes	No
3122	STAT3 Remaining Exons Sequencing	Yes	Yes	No
312	STAT3 Select Exons Sequencing	Yes	Yes	No
516	STRA6 Gene Sequencing	Yes	Yes	No
611	SUMF1 Gene Sequencing	Yes	Yes	No
494	TAT Gene Sequencing	Yes	Yes	No
358	TBX1 Gene Sequencing	Yes	Yes	No
653	TCOF1 Gene Sequencing	Yes	Yes	No
107	TERC Gene Sequencing	Yes	Yes	Yes
682	TERT Gene Sequencing	Yes	Yes	No
512	TGFBR1 Gene Sequencing	Yes	Yes	No
125	TGM1 Gene Sequencing	Yes	Yes	Yes
414	TINF2 Exon 6 Sequencing	Yes	Yes	Yes
555	TMEM127 Gene Sequencing	Yes	Yes	No
215	TNFRSF1A Select Exons Sequencing	Yes	Yes	Yes
559	TP53 Gene Sequencing	Yes	Yes	No
1581	TP63 Remaining Exons Sequencing	Yes	Yes	No
158	TP63 Select Exons Sequencing	Yes	Yes	No
489	TRPM1 Gene Sequencing	Yes	Yes	No
3275	TSPAN12 Gene Sequencing	Yes	Yes	Yes
374	UBE3A Gene Sequencing	Yes	Yes	No
670	UNG Gene Sequencing	Yes	Yes	No
908	Usher Syndrome Del/Dup Panel	Yes	No	No
585	Usher Syndrome Panel	Yes	No	No
413	VHL Gene Sequencing	Yes	Yes	Yes
2271	VPS13B (COH1) Targeted (Finnish mutation) Gene Sequencing	Yes	Yes	No
344	VSX2 (CHX10) Gene Sequencing	Yes	Yes	Yes
505	WAS Gene Sequencing	Yes	Yes	Yes
373	WNT10A Gene Sequencing	Yes	Yes	Yes
2571	XLRS1 (RS1) Gene Sequencing	Yes	Yes	Yes
502	ZRS Sequence Analysis (Intron 5 of LMBR1 Gene)	Yes	Yes	No
437	Zygoty Testing	Yes	No	Yes

Deletion/Duplication Testing

Test Code	Test Name	Buccal*	Oral Rinse	DBS
906	AAAS Del/Dup	Yes	Yes	No
906	ABCA12 Del/Dup	Yes	Yes	No
906	ABCA4 Del/Dup	Yes	Yes	No
906	ABCC6 Del/Dup	Yes	Yes	No
906	ABHD5 Del/Dup	Yes	Yes	No
906	ACAD8 Del/Dup	Yes	Yes	No
906	ACADM Del/Dup	Yes	Yes	No
906	ACADS Del/Dup	Yes	Yes	No
906	ACADSB Del/Dup	Yes	Yes	No
906	ACADVL Del/Dup	Yes	Yes	No

Deletion/Duplication Testing

Test Code	Test Name	Buccal*	Oral Rinse	DBS
906	ACAT1 Del/Dup	Yes	Yes	No
906	ACSF3 Del/Dup	Yes	Yes	No
906	ADA Del/Dup	Yes	Yes	No
906	AGA Del/Dup	Yes	Yes	No
906	AICDA Del/Dup	Yes	Yes	No
906	AIPL1 Del/Dup	Yes	Yes	No
906	AIRE Del/Dup	Yes	Yes	No
906	ALDH3A2 Del/Dup	Yes	Yes	No
906	ALOX12B Del/Dup	Yes	Yes	No
906	ALOXE3 Del/Dup	Yes	Yes	No
906	AR Del/Dup	Yes	Yes	No
906	ARG1 Del/Dup	Yes	Yes	No
906	ARSA Del/Dup	Yes	Yes	No
906	ARSB Del/Dup	Yes	Yes	No
906	ARSE Del/Dup	Yes	Yes	No
906	ARX Del/Dup	Yes	Yes	No
906	ASL Del/Dup	Yes	Yes	No
906	ASPA Del/Dup	Yes	Yes	No
906	ATP2A2 Del/Dup	Yes	Yes	No
906	AUH Del/Dup	Yes	Yes	No
906	BCKDHA Del/Dup	Yes	Yes	No
906	BCKDHB Del/Dup	Yes	Yes	No
906	BCOR Del/Dup	Yes	Yes	No
906	BEST1 Del/Dup	Yes	Yes	No
906	BLM Del/Dup	Yes	Yes	No
906	BRAF Del/Dup	Yes	Yes	No
906	BTD Del/Dup	Yes	Yes	No
906	BTK Del/Dup	Yes	Yes	No
906	CASP8 Del/Dup	Yes	Yes	No
906	CBS Del/Dup	Yes	Yes	No
906	CCM Del/Dup Panel	Yes	Yes	No
906	CCM2 Del/Dup	Yes	Yes	No
906	CD40LG Del/Dup	Yes	Yes	No
906	CDC73 (HRPT2) Del/Dup	Yes	Yes	No
906	CDKL5 Del/Dup	Yes	Yes	No
906	CEP290 Del/Dup	Yes	Yes	No
906	CERKL Del/Dup	Yes	Yes	No
906	CHD7 Del/Dup	Yes	Yes	No
906	CHM Del/Dup	Yes	Yes	No
906	CLCN5 Del/Dup (Females)	Yes	Yes	No
906	CNGA1 Del/Dup	Yes	Yes	No
906	CNGA3 Del/Dup	Yes	Yes	No
906	CNGB3 Del/Dup	Yes	Yes	No
906	COL17A1 Del/Dup	Yes	Yes	No
906	COL4A5 Del/Dup	Yes	Yes	No
906	COL7A1 Del/Dup	Yes	Yes	No

Deletion/Duplication Testing

Test Code	Test Name	Buccal*	Oral Rinse	DBS
906	COMP Del/Dup	Yes	Yes	No
906	CPT1A Del/Dup	Yes	Yes	No
906	CPT2 Del/Dup	Yes	Yes	No
906	CRB1 Del/Dup	Yes	Yes	No
906	CREBBP Del/Dup	Yes	Yes	No
906	CRX Del/Dup	Yes	Yes	No
906	CYBA Del/Dup	Yes	Yes	No
906	CYBB Del/Dup	Yes	Yes	No
906	CYP17A1 Del/Dup	Yes	Yes	No
906	CYP19A1 Del/Dup	Yes	Yes	No
906	CYP1B1 Del/Dup	Yes	Yes	No
906	DBT Del/Dup	Yes	Yes	No
906	DCLRE1C Del/Dup	Yes	Yes	No
904	Del/Dup (qPCR/MLPA) Testing	Yes	Yes	No
904	Del/Dup Current Menu Gene	Yes	Yes	No
906	DHCR7 Del/Dup	Yes	Yes	No
906	DHH Del/Dup	Yes	Yes	No
906	DKC1 Del/Dup	Yes	Yes	No
906	DLD Del/Dup	Yes	Yes	No
906	DOCK8 Del/Dup	Yes	Yes	No
906	EDA1 Del/Dup	Yes	Yes	No
906	EDAR Del/Dup	Yes	Yes	No
906	EFNB1 Del/Dup	Yes	Yes	No
906	ELOVL4 Del/Dup	Yes	Yes	No
906	ETFA Del/Dup	Yes	Yes	No
906	ETFB Del/Dup	Yes	Yes	No
906	ETFDH Del/Dup	Yes	Yes	No
906	ETHE1 Del/Dup	Yes	Yes	No
906	EYA1 Del/Dup	Yes	Yes	No
906	EYS Del/Dup	Yes	Yes	No
906	FAH Del/Dup	Yes	Yes	No
906	FH Del/Dup	Yes	Yes	No
906	FLCN Del/Dup	Yes	Yes	No
906	FOXC1 Del/Dup	Yes	Yes	No
906	FRMD7 Del/Dup (Females)	Yes	Yes	No
906	FUCA1 Del/Dup	Yes	Yes	No
906	FZD4 Del/Dup	Yes	Yes	No
906	GAA Del/Dup	Yes	Yes	No
906	GALC Del/Dup	Yes	Yes	No
906	GALK1 Del/Dup	Yes	Yes	No
906	GALNS Del/Dup	Yes	Yes	No
906	GALT Del/Dup	Yes	Yes	No
906	GCDH Del/Dup	Yes	Yes	No
906	GCH1 Del/Dup	Yes	Yes	No
906	GK Del/Dup	Yes	Yes	No
906	GLA Del/Dup (Females)	Yes	Yes	No

Deletion/Duplication Testing

Test Code	Test Name	Buccal*	Oral Rinse	DBS
906	GLB1 Del/Dup	Yes	Yes	No
906	GNE Del/Dup	Yes	Yes	No
906	GNS Del/Dup	Yes	Yes	No
906	GPC3 Del/Dup	Yes	Yes	No
906	GUCA1A Del/Dup	Yes	Yes	No
906	GUCY2D Del/Dup	Yes	Yes	No
906	HADH Del/Dup	Yes	Yes	No
906	HADHA Del/Dup	Yes	Yes	No
906	HADHB Del/Dup	Yes	Yes	No
906	HAX1 Del/Dup	Yes	Yes	No
906	HESX1 Del/Dup	Yes	Yes	No
906	HEXA Del/Dup	Yes	Yes	No
906	HEXB Del/Dup	Yes	Yes	No
906	HGSNAT Del/Dup	Yes	Yes	No
906	HLCS Del/Dup	Yes	Yes	No
906	HMGCL Del/Dup	Yes	Yes	No
906	HOXD13 Del/Dup	Yes	Yes	No
906	HPD Del/Dup	Yes	Yes	No
906	HRAS Del/Dup	Yes	Yes	No
906	HSD17B10 Del/Dup (Females)	Yes	Yes	No
906	IGHMBP2 Del/Dup	Yes	Yes	No
906	IL2RG Del/Dup	Yes	Yes	No
906	IL7R Del/Dup	Yes	Yes	No
906	IRAK4 Del/Dup	Yes	Yes	No
906	IRF6 Del/Dup	Yes	Yes	No
906	ITGB2 Del/Dup	Yes	Yes	No
906	ITGB4 Del/Dup	Yes	Yes	No
906	IVD Del/Dup	Yes	Yes	No
906	JAG1 Del/Dup	Yes	Yes	No
906	JAK3 Del/Dup	Yes	Yes	No
906	KAL1 Del/Dup	Yes	Yes	No
906	KMT2D Del/Dup	Yes	Yes	No
906	KRAS Del/Dup	Yes	Yes	No
906	L1CAM Del/Dup	Yes	Yes	No
906	LAMA3 Del/Dup	Yes	Yes	No
906	LAMB3 Del/Dup	Yes	Yes	No
906	LAMC2 Del/Dup	Yes	Yes	No
906	LIPA Del/Dup	Yes	Yes	No
906	LRP5 Del/Dup	Yes	Yes	No
906	MAP2K1 Del/Dup	Yes	Yes	No
906	MAP2K2 Del/Dup	Yes	Yes	No
906	MAT1A Del/Dup	Yes	Yes	No
906	MCCC1 Del/Dup	Yes	Yes	No
906	MCCC2 Del/Dup	Yes	Yes	No
906	MCOLN1 Del/Dup	Yes	Yes	No
904	MEN1 Del/Dup	Yes	Yes	No

Deletion/Duplication Testing

Test Code	Test Name	Buccal*	Oral Rinse	DBS
906	MLYCD Del/Dup	Yes	Yes	No
906	MMAA Del/Dup	Yes	Yes	No
906	MMAB Del/Dup	Yes	Yes	No
906	MMACHC Del/Dup	Yes	Yes	No
906	MPL Del/Dup	Yes	Yes	No
906	MUT Del/Dup	Yes	Yes	No
906	MVK Del/Dup	Yes	Yes	No
906	MYCN Del/Dup	Yes	Yes	No
906	MYOC Del/Dup	Yes	Yes	No
906	NAGLU Del/Dup	Yes	Yes	No
906	NAGS Del/Dup	Yes	Yes	No
906	NCF2 Del/Dup	Yes	Yes	No
906	NDP Del/Dup	Yes	Yes	No
906	NEU1 Del/Dup	Yes	Yes	No
906	NIPBL Del/Dup	Yes	Yes	No
906	NPC1 Del/Dup	Yes	Yes	No
906	NPC2 Del/Dup	Yes	Yes	No
906	NR0B1 (DAX1) Del/Dup	Yes	Yes	No
906	NR5A1 Del/Dup	Yes	Yes	No
906	NSD1 Del/Dup	Yes	Yes	No
906	NTRK1 Del/Dup	Yes	Yes	No
906	NYX Del/Dup (Females)	Yes	Yes	No
906	OCRL Del/Dup (Females)	Yes	Yes	No
906	OFD1 (CXORF5) Del/Dup	Yes	Yes	No
906	OPTN Del/Dup	Yes	Yes	No
906	OTC Del/Dup	Yes	Yes	No
906	OTX2 Del/Dup	Yes	Yes	No
906	PAH Del/Dup	Yes	Yes	No
906	PAX2 Del/Dup	Yes	Yes	No
906	PAX6 Del/Dup	Yes	Yes	No
906	PC Del/Dup	Yes	Yes	No
906	PCCA Del/Dup	Yes	Yes	No
906	PCCB Del/Dup	Yes	Yes	No
906	PDCD10 Del/Dup	Yes	Yes	No
906	PDE6B Del/Dup	Yes	Yes	No
906	PDHA1 Del/Dup (Females)	Yes	Yes	No
906	PDHB Del/Dup	Yes	Yes	No
906	PHEX Del/Dup	Yes	Yes	No
906	PITX2 Del/Dup	Yes	Yes	No
906	POLG Del/Dup	Yes	Yes	No
906	POR Del/Dup	Yes	Yes	No
906	PORCN Del/Dup	Yes	Yes	No
906	PRKAR1A Del/Dup	Yes	Yes	No
906	PRPF3 Del/Dup	Yes	Yes	No
906	PRPF31 Del/Dup	Yes	Yes	No
906	PRPF8Del/Dup	Yes	Yes	No

Deletion/Duplication Testing

Test Code	Test Name	Buccal*	Oral Rinse	DBS
906	PSAP Del/Dup	Yes	Yes	No
906	PTEN Del/Dup	Yes	Yes	No
906	PTPN11 Del/Dup	Yes	Yes	No
906	PTS Del/Dup	Yes	Yes	No
906	PUS1 Del/Dup	Yes	Yes	No
906	PYGM Del/Dup	Yes	Yes	No
906	QDPR Del/Dup	Yes	Yes	No
906	RAF1 Del/Dup	Yes	Yes	No
906	RAG1 Del/Dup	Yes	Yes	No
906	RAG2 Del/Dup	Yes	Yes	No
906	RAI1 Del/Dup	Yes	Yes	No
906	RDH5 Del/Dup	Yes	Yes	No
906	RDS (PRPH2) Del/Dup	Yes	Yes	No
906	RECQL4 Del/Dup	Yes	Yes	No
906	RET Del/Dup	Yes	Yes	No
906	RHO Del/Dup	Yes	Yes	No
906	RLBP1 Del/Dup	Yes	Yes	No
906	RMRP Del/Dup	Yes	Yes	No
906	RP1 Del/Dup	Yes	Yes	No
906	RP2 Del/Dup (Females)	Yes	Yes	No
906	RPE65 Del/Dup	Yes	Yes	No
906	RPS19 Del/Dup	Yes	Yes	No
906	RSK2 (RPS6KA3) Del/Dup	Yes	Yes	No
906	SALL1 Del/Dup	Yes	Yes	No
906	SALL4 Del/Dup	Yes	Yes	No
906	SDHB, SDHC & SDHD Del/Dup	Yes	Yes	No
906	SERPING1 (C11NH) Del/Dup	Yes	Yes	No
906	SGSH Del/Dup	Yes	Yes	No
906	SHH Del/Dup	Yes	Yes	No
906	SIX1 Del/Dup	Yes	Yes	No
906	SIX3 Del/Dup	Yes	Yes	No
906	SIX6 Del/Dup	Yes	Yes	No
906	SLC17A5 Del/Dup	Yes	Yes	No
906	SLC22A5 Del/Dup	Yes	Yes	No
906	SLC25A13 Del/Dup	Yes	Yes	No
906	SLC25A20 Del/Dup	Yes	Yes	No
906	SMC1A Del/Dup	Yes	Yes	No
906	SMPD1 Del/Dup	Yes	Yes	No
906	SOX2 Del/Dup	Yes	Yes	No
906	SOX9 Del/Dup	Yes	Yes	No
906	SPINK5 Del/Dup	Yes	Yes	No
906	SPRED1 Del/Dup	Yes	Yes	No
906	SRD5A2 Del/Dup	Yes	Yes	No
906	SUMF1 Del/Dup	Yes	Yes	No
906	TAT Del/Dup	Yes	Yes	No
904	TBX1 Del/Dup	Yes	Yes	No

Deletion/Duplication Testing

Test Code	Test Name	Buccal*	Oral Rinse	DBS
906	TBX5 Del/Dup	Yes	Yes	No
906	TCOF1 Del/Dup	Yes	Yes	No
906	TERC Del/Dup	Yes	Yes	No
906	TGM1 Del/Dup	Yes	Yes	No
906	TH Del/Dup	Yes	Yes	No
906	TINF2 Del/Dup	Yes	Yes	No
906	TP53 Del/Dup	Yes	Yes	No
906	TP63 Del/Dup	Yes	Yes	No
906	TPP1 Del/Dup	Yes	Yes	No
906	TTR Del/Dup	Yes	Yes	No
906	UBE3A Del/Dup	Yes	Yes	No
906	VPS13B (COH1) Del/Dup	Yes	Yes	No
906	WAS Del/Dup	Yes	Yes	No
906	WNT10A Del/Dup	Yes	Yes	No
906	XLR1 (RS1) Del/Dup (Females)	Yes	Yes	No
906	ZIC2 Del/Dup	Yes	Yes	No
906	ZRS Del/Dup Analysis (Intron 5 of LMBR1 Gene)	Yes	Yes	No

*denotes GeneDx Sponge Swab



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