Immunology Exome

Genes Tested:

ACD, ACP5, ACTB, ADA, ADA2, ADAM17, ADAR, AICDA, AIRE, AK2, ALPI, AP1S3, AP3B1, AP3D1, APOL1, ARHGEF1, ARPC1B, ATM, ATP6AP1, B2M, BACH2, BCL10, BCL11B, BLM, BLNK, BRCA1, BRCA2, BRIP1, BTK, C1QA, C1QB, C1QC, C1R, C1S, C2, C3, C4BPA, C5, C6, C7, C8A, C8B, C8G, C9, CARD11, CARD14, CARD9, CARMIL2, CASP10, CASP8, CCBE1, CD19, CD247, CD27, CD3D, CD3E, CD3G, CD40, CD40LG, CD46, CD55, CD59, CD70, CD79A, CD79B, CD81, CD8A, CDC42, CDCA7, CEBPE, CFB, CFD, CFH, CFHR1, CFHR2, CFHR3, CFHR5, CFI, CFP, CFTR, CHD7, CIB1, CIITA, CLCN7, CLPB, COG6, COPA, CORO1A, CR2, CREBBP, CSF2RA, CSF2RB, CSF3R, CTC1, CTLA4, CTPS1, CTSC, CXCR4, CYBA, CYBB, DBR1, DCLRE1B, DCLRE1C, DDX58, DEF6, DGKE, DKC1, DNAJC21, DNASE1L3, DNASE2, DNMT3B, DOCK2, DOCK8, DSG1, EFL1, ELANE, EPG5, ERBIN, ERCC2, ERCC3, ERCC4, ERCC6L2, EXTL3, FAAP24, FADD, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FAS, FASLG, FAT4, FCGR3A, FCHO1, FCN3, FERMT1, FERMT3, FOXN1, FOXP3, FPR1, G6PC, G6PC3, G6PD, GATA1, GATA2, GFI1, GINS1, GTF2H5, HAVCR2, HAX1, HELLS, HMOX1, HYOU1, ICOS, ICOSLG, IFIH1, IFNAR1, IFNAR2, IFNGR1, IFNGR2, IGLL1, IKBKB, IKZF1, IL10, IL10RA, IL10RB, IL12B, IL12RB1, IL12RB2, IL17F, IL17RA, IL17RC, IL18BP, IL1RN, IL21, IL21R, IL23R, IL2RA, IL2RB, IL2RG, IL36RN, IL6R, IL6ST, IL7R, INO80, INSR, IRAK1, IRAK4, IRF2BP2, IRF3, IRF7, IRF4, IRF8, IRF9, ISG15, ITCH, ITGAM, ITGB2, ITK, JAGN1, JAK1, JAK3, KDM6A, KMT2A, KMT2D, KRAS, LAMTOR2, LAT, LCK, LIG1, LIG4, LPIN2, LRBA, LRRC8A, LYST, MAD2L2, MAGT1, MALT1, MAN2B1, MAP3K14, MASP2, MBL2, MCM4, MEFV, MKL1, MOGS, MPO, MRE11, MS4A1, MSH6, MSN, MTHFD1, MVK, MYD88, MYH9, MYO5A, MYSM1, NBAS, NBN, NCF2, NCF4, NCSTN, NFAT5, NFE2L2, NFKB1, NFKB2, NFKBIA, NHEJ1, NHP2, NLRC4, NLRP1, NLRP12, NLRP3, NOD2, NOP10, NRAS, NSMCE3, OAS1, OFD1, ORAI1, OSTM1, OTULIN, PALB2, PARN, PCCA, PCCB, PEPD, PGM3, PIGA, PIK3CD, PIK3R1, PLCG2, PLEKHM1, PNP, POLA1, POLD1, POLD2, POLE, POLE2, POLR3A, POLR3C, POLR3F, PRF1, PRKCD, PRKDC, PSENEN, PSMA3, PSMB4, PSMB8, PSMG2, PSTPIP1, PTEN, PTPRC, RAB27A, RAC2, RAD50, RAD51, RAD51C, RAG1, RAG2, RASGRP1, RBCK1, RBM8A, RECQL4, REL, RELA, RELB, RFWD3, RFX5, RFXANK, RFXAP, RHOH, RIPK1, RNASEH2A, RNASEH2B, RNASEH2C, RNF168, RNF31, RORC, RPL11, RPL15, RPL26, RPL35A, RPL36, RPL5, RPS10, RPS15, RPS15A, RPS19, RPS24, RPS26, RPS27A, RPS28, RPS29, RPS7, RPSA, RTEL1, RUNX1, SAMD9, SAMD9L, SAMHD1, SBDS, SEC61A1, SEMA3E, SERPING1, SH2D1A, SH3BP2, SH3KBP1, SKIV2L, SLC29A3, SLC35A1, SLC35C1, SLC37A4, SLC39A4, SLC39A7, SLC46A1, SLC7A7, SLX4, SMARCAL1, SMARCD2, SNX10, SP110, SPINK5, SPPL2A, SRP54, SRP72, STAT1, STAT2, STAT3, STAT5B, STIM1, STK4, STN1, STX11, STXBP2, TAP1, TAP2, TAPBP, TAZ, TBK1, TBX1, TCF3, TCIRG1, TCN2, TERT, TFRC, TGFB1, TGFBR1, TGFBR2, THBD, TICAM1, TINF2, TIRAP, TLR3, TLR8, TMC6, TMC8, TMEM173, TNFAIP3, TNFRSF11A, TNFRSF13B, TNFRSF13C, TNFRSF1A, TNFRSF4, TNFRSF9, TNFSF11, TNFSF12, TOP2B, TP53, TPP2, TRADD, TRAF3, TRAF3IP2, TREX1, TRIM22, TRNT1, TTC37, TTC7A, TYK2, UBE2T, UNC13D, UNC93B1, UNG, USB1, VPS13B, VPS45, WAS, WDR1, WIPF1, WRAP53, XIAP, XK, XRCC2, XRCC4, ZAP70, ZBTB24, ZNF341

Note: *C4A, C4B, NCF1, IKBKG (NEMO), RANBP2, USP18, RPS17, TERC are not analyzed in the immunology exome due to high homology or lack of coverage.*

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Genetics and Genomics Diagnostic Laboratory
CLIA#: 36D0656333
Phone: (513) 636-4474
Fax: (513) 636-4373
Email: LabGeneticCounselors@cchmc.org
www.cincinnatichildrens.org/moleculargenetics

Description:

Our Immunology Exome utilizes Whole Exome Sequencing (WES) technology but focuses on a predefined list of 442 genes that are associated with immune system defects or related disorders. The genes included in this test are associated with susceptibility to recurrent or unusual infections, antibody deficiencies, immune dysregulation, malignancy, allergy, autoimmunity, and auto-inflammatory disorders. Our extensive gene list was developed through careful review of available evidence and collaboration with clinical immunologists and researchers. Compared to WES, this targeted approach results in a shorter turnaround time and decreased cost. This test will be performed on the proband only and will not include the identification of ACMG recommended actionable incidental findings.

Indications:

- Immunodeficiencies affecting cellular and humoral immunity
- Combined immunodeficiencies with associated or syndromic features
- Predominantly antibody deficiencies
- Diseases of immune dysregulation
- Congenital defects of phagocyte number or function
- Defects in intrinsic and innate immunity
- Autoinflammatory disorders
- Complement deficiencies
- Bone marrow failure

Over 370 of the 442 genes in our Immunology Exome have been classified in the Human Inborn Errors of Immunity: 2019 Update on the Classification from the International Union of Immunological Societies Expert Committee. Their phenotypic classifications (published in J Clin Immunol. 40(1): 24–64.) are outlined below:

2019 IUIS Table 1: Immunodeficiencies affecting cellular and humoral immunity

ADA, AK2, B2M, BCL10, BCL11B, CARD11, CD247, CD3D, CD3E, CD3G, CD40, CD40LG, CD8A, CDC42, CIITA, CORO1A, DCLRE1C, DOCK2, DOCK8, FCHO1, ICOS, ICOSLG, IKBKB, IL21, IL21R, IL2RG, IL7R, JAK3, LAT, LCK, LIG4, MALT1, MAP3K14, MSN, NHEJ1, POLD1, POLD2, PRKDC, PTPRC, RAG1, RAG2, REL, RELA, RELB, RFX5, RFXANK, RFXAP, RHOH, STK4, TAP1, TAP2, TAPBP, TFRC, TLR8, TNFRSF4, ZAP70

2019 IUIS Table 2: Combined immunodeficiencies with associated or syndromic features

ACD, ARPC1B, ATM, BLM, CCBE1, CDCA7, CHD7, CTC1, DCLRE1B, DKC1, DNMT3B, EPG5, ERCC6L2, ERBIN, EXTL3, FAT4, FOXN1, GINS1, HELLS, IL6R, IL6ST, KDM6A, KMT2A, KMT2D, LIG1, MCM4, MTHFD1, MYSM1, NBN, NFE2L2, NFKBIA, NHP2, NOP10, NSMCE3, ORAI1, PARN, PGM3, PNP, POLE, POLE2, RBCK1, RNF168, RNF31, RTEL1, SAMD9, SAMD9L, SEMA3E, SLC46A1, SMARCAL1, SP110, SPINK5, STAT3, STAT5B, STIM1, STN1, TBX1, TCN2, TERT, TGFBR1, TGFBR2, TINF2, TTC7A, WAS, WIPF1, WRAP53, ZBTB24, ZNF341

2019 IUIS Table 3: Predominantly antibody deficiencies

CDA, ARHGEF1, ATP6AP1, BLNK, BTK, CARD11, CD19, CD79A, CD79B, CD81, CR2, IGLL1, IKZF1, INO80, IRF2BP2, MOGS, MS4A1, MSH6, NFKB1, NFKB2, PIK3CD, PIK3R1, PTEN, SEC61A1, SH3KBP1, SLC39A7, TCF3, TNFRSF13B, TNFRSF13C, TNFSF12, TOP2B, TRNT1, TTC37, UNG

2019 IUIS Table 4: Diseases of immune dysregulation

AIRE, AP3B1, AP3D1, BACH2, CARMIL2, CASP10, CASP8, CD27, CD70, CTLA4, CTPS1, DEF6, FAAP24, FADD, FAS, FASLG, FERMT1, FOXP3, IL10, IL10RA, IL10RB, IL2RA, IL2RB, ITCH, ITK, JAK1, LRBA, LYST, MAGT1, NFAT5, PEPD, PRF1, PRKCD, RAB27A, RASGRP1, RIPK1, SH2D1A, STAT3, STX11, STXBP2, TGFB1, TNFRSF9, TPP2, UNC13D, XIAP, ZAP70

2019 IUIS Table 5: Congenital defects of phagocyte number or function

ACTB, CEBPE, CFTR, CLPB, CSF2RA, CSF2RB, CSF3R, CTSC, CYBA, CYBB, DNAJC21, EFL1, ELANE, FERMT3, FPR1, G6PC, G6PC3, G6PD, GATA2, GFI1, HAX1, HYOU1, ITGB2, JAGN1, LAMTOR2, MKL1, NCF2, NCF4, RAC2, SBDS, SLC35C1, SLC37A4, SMARCD2, TAZ, USB1, VPS13B, VPS45, WAS, WDR1

2019 IUIS Table 6: Defects in intrinsic and innate immunity

APOL1, CARD9, CLCN7, CIB1, CXCR4, CYBB, DBR1, FCGR3A, HMOX1, IFIH1, IFNAR1, IFNAR2, IFNGR1, IFNGR2, IL12B, IL12RB1, IL12RB2, IL17F, IL17RA, IL17RC, IL18BP, IL23R, IRAK1, IRAK4, IRF4, IRF3, IRF7, IRF8, IRF9, ISG15, JAK1, MYD88, NBAS, NCSTN, OSTM1, PLEKHM1, POLR3A, POLR3C, POLR3F, PSENEN, RORC, RPSA, SNX10, SPPL2A, STAT1, STAT2, TBK1, TCIRG1, TICAM1, TIRAP, TLR3, TMC6, TMC8, TNFRSF11A, TNFSF11, TRAF3, TRAF3IP2, TYK2, UNC93B1

2019 IUIS Table 7: Autoinflammatory disorders

ACP5, ADA2, ADAM17, ADAR, ALPI, AP1S3, CARD14, COPA, DDX58, HAVCR2, IFIH1, IL1RN, IL36RN, LPIN2, MEFV, MVK, NLRC4, NLRP1, NLRP12, NLRP3, NOD2, OAS1, OTULIN, PLCG2, POLA1, PSMA3, PSMB4, PSMB8, PSMG2, PSTPIP1, RNASEH2A, RNASEH2B, RNASEH2C, SAMHD1, SH3BP2, SLC29A3, TMEM173, TNFAIP3, TNFRSF1A, TREX1, TRIM22

2019 IUIS Table 8: Complement deficiencies
C1QA, C1QB, C1QC, C1R, C1S, C2, C3, C5, C6, C7, C8A, C8B, C8G, C9, CD46, CD55, CD59, CFB, CFD, CFH, CFHR1, CFHR2, CFHR3, CFHR5, CFI, CFP, FCN3, MASP2, SERPING1, THBD

2019 IUIS Table 9: Bone marrow failure
AIRE, BRCA1, KRAS, MAD2L2, NLRP3, NRAS, RAD51, RFWD3, STAT3, STAT5B, TP53, UBE2T, XRCC2

Additional 68 genes related to inherited immunodeficiencies are also analyzed in our Immunology Exome:

Additional clinically relevant genes
BRCA2, BRIP1, C4BPA, COG6, CREBBP, DGKE, DNASE1L3, DNASE2, DSG1, ERCC2, ERCC3, ERCC4, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, GATA1, GTF2H5, INSR, ITGAM, LRRC8A, MAN2B1, MBL2, MPO, MRE11, MYH9, MYO5A, OFD1, PALB2, PCCA, PCCB, PIGA, RAD50, RAD51C, RBM8A, RECQL4, RPL11, RPL15, RPL26, RPL35A, RPL36, RPL5, RPS10, RPS15, RPS15A, RPS19, RPS24, RPS26, RPS27A, RPS28, RPS29, RPS7, RUNX1, SKIV2L, SLC35A1, SLC39A4, SLC7A7, SLX4, SRP54, SRP72, TRADD, XK, XRCC4

What Is Reported?

Variants that will be discussed in detail in the report:

- **Pathogenic/likely pathogenic variants:** Variants that are known to be pathogenic or for whichthe laboratory has sufficient evidence suggesting pathogenicity.

Variants that will be listed in the report:

- Variants of uncertain clinical significance.

What is not reported?

- Variants in genes not included in the predefined gene list
- Variants where there is currently no evidence of association with the disease and that are identified in healthy individuals (benign or likely benign variants)
- Variants that predict an increased risk of diseases, but do not cause a disease by themselves (risk alleles).

Note: Immunology Exome cases with negative or uncertain findings can be reflexed to Whole Exome Sequencing (WES). A separate test order and a signed consent form is required for all WES testing. In addition, including biological parental samples is strongly encouraged to assist with the analysis of WES and to increase test yield. Reflex to WES orders can either be placed simultaneously or separately. Separate reflex to WES orders are subject to review prior to the initiation of testing. Please see our website at www.cincinnatichildrens.org/exome to obtain a WES test requisition and consent form.

Methodology:

Immunology Exome uses the Human Comprehensive Exome kit to capture the exonic regions of genes from the genomic DNA extracted from the patient. Targeted regions are sequenced using an Illumina sequencing system with paired-end reads. Sequence reads are aligned to the human reference genome (build UCSC hg19). Variants within exons and flanking sequences are identified and evaluated by a validated in-house developed bioinformatics analysis pipeline and Fabric Genomic Analysis platform. Allele specific analysis for the 253kb inversion as well as targeted analysis of the c.118-308 region in UNC13D are performed. Data quality is assessed to confirm it has a minimum coverage of 20X for >95% of targets of interest.

Technical Limitations:

- Pathogenic variants may be present in a portion of the genes not covered by this test or in regions with suboptimal data due to homologous issue, polynucleotides, or nucleotide repeats, and therefore may not be identified. Thus, the absence of identified pathogenic variants does not exclude the possibility of a genetic etiology for the patient's symptoms.
- Certain types of mutations are not detected. Only single base pair changes or small insertions or deletions of DNA are detected. Large deletions, duplications, or rearrangements, mitochondrial genome mutations, repeat expansions, low level mosaicism and many epigenetic defects may not be detected by this test.

Low coverage (<20X) regions

GENE	TRANSCRIPT	EXON	CHROM	EXON_START	EXON_END
CD55	NM_001300902	10	1	207513735	207513989
CD55	NM_001300903	10	1	207520791	207520906
IL12RB1	NM_153701	10	19	18182018	18182143
IL6R	NM_001206866	7	1	154409910	154410020
RAD51	NM_133487	4	15	40994003	40994124
VPS13B	NM_015243	18	8	100221819	100221896
VPS45	NM_001279353	13	1	150115015	150115109

Please note: *These regions represent the low coverage (<20X) regions identified during our test validation. For specific patient cases, these regions may vary slightly.*

Regions of Homology

These gene regions with homology may generate suboptimal data with potential false negative results.

GENE	TRANSCRIPT	EXON	CHROM	EXON_START	EXON_END
CDC42	NM_001791	6	1	22417920	22418010
CFHR1	NM_002113	1	1	196788974	196789032
CFHR2	NM_005666	1	1	196913010	196913068
CFHR3	NM_021023	4	1	196757345	196757528
CFHR3	NM_021023	5	1	196759174	196759357
CORO1A	NM_007074	11	16	30200180	30200285
EFL1	NM_024580	7	15	82530647	82530862
FANCD2	NM_033084	14	3	10085512	10085548
FANCD2	NM_033084	17	3	10091057	10091189
FANCD2	NM_033084	22	3	10106039	10106113
IGLL1	NM_152855	2	22	23915723	23915772
PLEKHM1	NM_014798	2	17	43559802	43559850
PLEKHM1	NM_014798	8	17	43527983	43528129
PLEKHM1	NM_014798	9	17	43522835	43523029
RBM8A	NM_005105	6	1	145509165	145509211
RPL15	NM_002948	4	3	23960686	23960992
RPSA	NM_002295	4	3	39452244	39452490
RPSA	NM_002295	7	3	39453764	39453859
STAT5B	NM_012448	7	17	40371329	40371481
STAT5B	NM_012448	8	17	40370740	40370896
UNC93B1	NM_030930	11	11	67761146	67761265
UNC93B1	NM_030930	12	11	67759016	67759328

Note: Targeted deletion and duplication analysis of every gene on this panel except ACD, ACP5, ACTB, ADAM17, ADAR, AICDA, AIRE, ALPI, AP1S3, APOL1, ARHGEF1, ATP6AP1, B2M, BACH2, BCL10, BLNK, BTK, C1QA, C1QB, C1QC, C1R, C1S, C2, C6, C7, C8A, C8B, C8G, C9, CARD11, CARD14,CARD9, CARMIL2, CCBE1, CD19, CD3G, CD40, CD70, CD79A, CD79B, CD81, CD8A, CDC42, CDCA7, CEBPE, CFHR1, CFHR2, CFHR3, CFTR, CIB1, CLCN7, COG6, COPA, CORO1A, CR2, CREBBP, CSF2RA, CSF2RB, CTSC, CYBA, CYBB, DBR1, DCLRE1B, DDX58, DEF6, DNASE1L3, DNASE2, DNMT3B, DOCK2, DSG1, EFL1, EPG5, ERBIN, ERCC2, ERCC3, EXTL3, FAAP24, FAT4, FCGR3A, FCHO1, FCN3, FERMT1, FERMT3, FPR1, GINS1, GTF2H5, HAVCR2, HELLS, HMOX1, HYOU1, ICOS, ICOSLG, IFIH1, IFNAR1, IFNAR2, IFNGR1, IFNGR2, IGLL1, IKBKB, IKZF1, IL10, IL10RA, IL10RB, IL12B, IL12RB1, IL12RB2, IL17F, IL17RA, IL17RC, IL18BP, IL1RN, IL21, IL21R, IL23R, IL2RA, IL2RB, IL36RN, IL6R, IL6ST, INO80, INSR, IRAK1, IRAK4, IRF2BP2, IRF3, IRF4, IRF7, IRF8, IRF9, ISG15, ITCH, ITGAM, ITGB2, JAK1, KDM6A, KMT2A, KMT2D, LCK, LIG1, LIG4, LRRC8A, MAD2L2, MALT1, MAN2B1, MAP3K14, MASP2, MBL2, MCM4, MEFV, MKL1, MOGS, MPO, MRE11, MS4A1, MSH6, MTHFD1, MVK, MYD88, MYO5A, NBAS, NCF2, NCF4, NCSTN, NFAT5, NFE2L2, NFKB1, NFKB2, NFKBIA, NHEJ1, NHP2, NLRC4, NLRP1, NLRP12, NLRP3, NOD2, NOP10, OAS1, OFD1, OSTM1, OTULIN, PCCA, PCCB, PEPD, PGM3, PIGA, PIK3R1, PLCG2, PLEKHM1, POLA1, POLD1, POLD2, POLE, POLE2, POLR3A, POLR3C, POLR3F, PRKCD, PSENEN, PSMA3, PSMB4, PSMB8, PSMG2, PSTPIP1, PTEN, RAD50, RBCK1, RECQL4, REL, RELA, RELB, RFWD3, RHOH, RIPK1, RNASEH2A, RNASEH2B, RNASEH2C, RNF31, RORC, RPL15, RPL36, RPS15A, RPS24, RPS28, RPS29, RPSA, SAMD9, SAMD9L, SAMHD1, SEC61A1, SEMA3E, SERPING1, SH3BP2, SH3KBP1, SKIV2L, SLC29A3, SLC35A1, SLC35C1, SLC37A4, SLC39A4, SLC39A7, SLC46A1, SMARCAL1, SMARCD2, SNX10, SP110, SPINK5, SPPL2A, SRP54, STAT1, STAT2, TAPBP, TBK1, TCF3, TFRC, TGFB1, TGFBR1, TGFBR2, TICAM1, TINF2, TIRAP, TLR3, TLR8, TMC6, TMC8, TMEM173, TNFAIP3, TNFRSF11A, TNFRSF13B, TNFRSF13C, TNFRSF1A, TNFRSF4, TNFRSF9, TNFSF11, TNFSF12, TOP2B, TPP2, TRADD, TRAF3, TRAF3IP2, TREX1, TRIM22, TRNT1, TTC37, TYK2, UNC93B1, UNG, XRCC4, ZBTB24, and ZNF341 is clinically available at an additional charge.

Turn-Around Time:

56 days (8 weeks)

Specimen:

At least 3 mls whole blood in a lavender top (EDTA) tube or saliva in an Oragene saliva kit. Please call 513-636-4474 for a free saliva collection kit. Label the tube with the patient's name, birth date, and date of collection. Alternatively, 10 mcg of high quality DNA may be submitted.

CPT Codes:

- Immunology Exome: **81405, 81406, 81407**
- Deletion and duplication analysis of any single gene on the Immunology Exome except *AIRE, ATM, BRCA1, BRCA2, BTK, CD40LG, CHD7, CFTR, CREBBP, G6PC, G6PD, IL2RG, KRAS, MEFV, PALB2, PCCA, PCCB, PTEN, RPS19, SLC37A4, STAT3, TAZ, TGFBR1, TGFBR2, TP53, VPS13B,* and *WAS*: **81479**
- Deletion and duplication analysis of *CD40LG* and *MEFV*: **81404**
- Deletion and duplication analysis of *IL2RG, KRAS, RPS19, STAT3, TGFBR1, TGFBR2,* and *TP53*: **81405**
- Deletion and duplication analysis of *AIRE, BTK, PALB2, PCCA, PCCB, SLC37A4, TAZ,* and *WAS*: **81406**
- Deletion and duplication analysis of *CHD7* and *CREBBP*: **81407**
- Deletion and duplication analysis of *ATM* and *VPS13B*: **81408**
- Deletion and duplication analysis of *BRCA1*: **81165**
- Deletion and duplication analysis of *BRCA2*: **81216**
- Deletion and duplication analysis of *CFTR*: **81222**
- Deletion and duplication analysis of *G6PD*: **81249**
- Deletion and duplication analysis of *G6PC*: **81250**
- Deletion and duplication analysis of *PTEN*: **81323**

Shipping Instructions:

Please enclose test **requisition** with sample. **All information must be completed before sample can be processed.**

Place samples in styrofoam mailer and ship at room temperature by overnight Federal Express to arrive Monday through Saturday.

Ship to:

Genetics and Genomics Diagnostic Laboratory
3333 Burnet Avenue NRB 1042
Cincinnati, OH 45229
513-636-4474

References:

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