

# PRIMARY IMMUNODEFICIENCIES TESTING REQUISITION

**All Information Must Be Completed Before Sample Can Be Processed**

## PATIENT INFORMATION

Patient Name: \_\_\_\_\_, \_\_\_\_\_, \_\_\_\_\_  
Last First MI

Address: \_\_\_\_\_  
\_\_\_\_\_

Home Phone: \_\_\_\_\_

MR# \_\_\_\_\_ Date of Birth \_\_\_\_\_ / \_\_\_\_\_ / \_\_\_\_\_

Gender: ☐ Male ☐ Female

## ETHNIC/RACIAL BACKGROUND (Choose All)

☐ European American (White) ☐ African-American (Black)

☐ Native American or Alaskan ☐ Asian-American

☐ Pacific Islander ☐ Ashkenazi Jewish ancestry

☐ Latino-Hispanic \_\_\_\_\_  
(specify country/region of origin)

☐ Other \_\_\_\_\_  
(specify country/region of origin)

## BILLING INFORMATION (Choose ONE method of payment)

### ☐ REFERRING INSTITUTION

Institution: \_\_\_\_\_

Address: \_\_\_\_\_

City/State/Zip: \_\_\_\_\_

Accounts Payable Contact Name: \_\_\_\_\_

Phone: \_\_\_\_\_

Fax: \_\_\_\_\_

Email: \_\_\_\_\_

### ☐ COMMERCIAL INSURANCE\*

**Insurance can only be billed if requested at the time of service.**

Policy Holder Name: \_\_\_\_\_

Gender: \_\_\_\_\_ Date of Birth \_\_\_\_\_ / \_\_\_\_\_ / \_\_\_\_\_

Authorization Number: \_\_\_\_\_

Insurance ID Number: \_\_\_\_\_

Insurance Name: \_\_\_\_\_

Insurance Address: \_\_\_\_\_

City/State/Zip: \_\_\_\_\_

Insurance Phone Number: \_\_\_\_\_

#### \* PLEASE NOTE:

- We will not bill Medicaid, Medicaid HMO, or Medicare except for the following: CCHMC Patients, CCHMC Providers, or Designated Regional Counties.
- If you have questions, please call 1-866-450-4198 for complete details.

## SAMPLE/SPECIMEN INFORMATION

**Has patient received a bone marrow transplant?** ☐ Yes ☐ No

**Note:** For post-transplant patients, we accept pre-transplant samples or post-transplant skin fibroblasts **ONLY** (blood, saliva, and cytobrushes are not accepted). Culturing of skin fibroblasts is done at an additional charge.

SPECIMEN TYPE: ☐ Amniotic fluid ☐ Blood ☐ Cytobrushes  
☐ Cord blood ☐ CVS ☐ Bone marrow ☐ Saliva  
☐ Tissue (specify): \_\_\_\_\_

Specimen Date: \_\_\_\_\_ / \_\_\_\_\_ / \_\_\_\_\_ Time: \_\_\_\_\_

Specimen Amount: \_\_\_\_\_

**Each test requires 3 mL of whole blood in EDTA tube. Please call before sending alternate tissue samples, and for free cytobrush or saliva collection kits.**

DRAWN BY: \_\_\_\_\_

\*Phlebotomist must initial tube of specimen to confirm sample identity

## REFERRING PHYSICIAN

Physician Name (print): \_\_\_\_\_

Address: \_\_\_\_\_

Phone: ( \_\_\_\_\_ ) \_\_\_\_\_ Fax: ( \_\_\_\_\_ ) \_\_\_\_\_

Email: \_\_\_\_\_

Genetic Counselor/Lab Contact Name: \_\_\_\_\_

Phone: ( \_\_\_\_\_ ) \_\_\_\_\_ Fax: ( \_\_\_\_\_ ) \_\_\_\_\_

Email: \_\_\_\_\_

\_\_\_\_\_ Date: \_\_\_\_/\_\_\_\_/\_\_\_\_

**Referring Physician Signature (REQUIRED)**

☐ Patient signed completed ABN

Medical Necessity Regulations: At the government's request, the Molecular Genetics Laboratories would like to remind all physicians that when ordering tests that will be paid under federal health care programs, including Medicare and Medicaid programs, that these programs will pay only for those tests the relevant program deems to be (1) included as covered services, (2) reasonable, (3) medically necessary for the treatment and diagnosis of the patient, and (4) not for screening purposes.

## INDICATIONS/DIAGNOSIS/ICD-9 CODE

Reason for Testing:

- ☐ Diagnostic testing in suspected affected patient
- ☐ Carrier testing

- ☐ Prenatal diagnosis (by previous arrangement only)

## TEST(S) REQUESTED

### Primary Immunodeficiency (Comprehensive testing)

#### ☐ Immunology Exome

394 gene panel utilizing Whole Exome Sequencing (WES) technology\*

- ☐ Reflex to deletion/duplication of single gene(s) (specify): \_\_\_\_\_

\*See page 5 for comprehensive gene list

### Autoimmune lymphoproliferative syndrome

- ☐ Autoimmune Lymphoproliferative Syndrome (ALPS) Panel by next generation sequencing (NGS)  
(*ADA2 (CECR1), CASP8, CASP10, CTLA4, FADD, FAS, FASLG, ITK, KRAS, LRBA, MAGT1, NRAS, PRKCD, RASGRP1, STAT3*)
- ☐ Reflex to deletion/duplication of all available genes on panel\*
- ☐ Reflex to deletion/duplication of single gene(s) (specify): \_\_\_\_\_

#### ☐ FAS (*TNFRSF6*)

- ☐ Reflex to deletion/duplication of FAS (*TNFRSF6*)

#### ☐ FASLG (*TNFSF6*)

- ☐ Reflex to deletion/duplication of FASLG (*TNFSF6*)

#### ☐ CASP10

- ☐ Reflex to deletion/duplication of CASP10

- ☐ Somatic FAS sequence analysis of sorted double-negative T cell (DNTC)  
(You MUST call 513-636-2731 in advance for specimen requirements and to schedule this test)

### Bone marrow failure syndromes

- ☐ Bone Marrow Failure Syndromes Panel by next-generation sequencing (NGS)  
(*ABCB7, ACD, ADA2 (CECR1), AK2, AP3B1, ATM, ATR, BLM, BRCA1, BRCA2, BRIP1, CD40LG, CLPB, CSF3R, CTC1, CXCR2, CXCR4, DKC1, DNAJC21, EFL1, EIF2AK3, ELANE, EPO, ERCC4, ERCC6L2, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, G6PC3, GATA1, GATA2, GFI1, HAX1, HYOU1, JAGN1, LAMTOR2, LIG4, LYST, MAD2L2, MPL, MRTFA (MKL1), MYSM1, NAF1, NBN, NHEJ1, NHP2, NOP10, NSMCE3, PALB2, PARN, POT1, RAB27A, RAC2, RAD51, RAD51C, RBM8A, RFW3, RMRP, RNF168, RPL11, RPL15, RPL18, RPL26, RPL27, RPL31, RPL35, RPL35A, RPL5, RPL9, RPS10, RPS15, RPS15A, RPS17, RPS19, RPS24, RPS26, RPS27, RPS27A, RPS28, RPS29, RPS7, RTEL1, RUNX1, SBDS, SLC37A4, SLX4, SMARCD2, SRP54, SRP72, STK4, STN1, TAZ, TCIRG1, TCN2, TERC, TERF2IP, TERT, TINF2, TP53, TSR2, UBE2T, USB1, VPS13B, VPS45, WAS, WDR1, WIPF1, WRAP53, XRCC2*)
- ☐ Reflex to deletion/duplication of all available genes on panel\*
- ☐ Reflex to deletion/duplication of single gene(s) (specify): \_\_\_\_\_

- ☐ SBDS gene sequencing for Shwachman Diamond syndrome

### ☐ Chromosome Breakage Disorders Panel by next-generation sequencing (NGS)

(*ATM, BLM, BRCA1, BRCA2, BRIP1, ERCC4, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, LIG4, MAD2L2, MYSM1, NBN, NHEJ1, NSMCE3, PALB2, RAD51, RAD51C, RFW3, SLX4, UBE2T, XRCC2*)

- ☐ Reflex to deletion/duplication of all available genes on panel\*

- ☐ Reflex to deletion/duplication of single gene(s) (specify): \_\_\_\_\_

### ☐ Diamond-Blackfan Anemia Panel by next-generation sequencing (NGS)

(*EPO, GATA1, RPL11, RPL15, RPL18, RPL26, RPL27, RPL31, RPL35, RPL35A, RPL5, RPL9, RPS10, RPS15, RPS15A, RPS17, RPS19, RPS24, RPS26, RPS27, RPS27A, RPS28, RPS29, RPS7, TSR2*)

- ☐ Reflex to deletion/duplication of all available genes on panel\*

- ☐ Reflex to deletion/duplication of single gene(s) (specify): \_\_\_\_\_

### ☐ Dyskeratosis Congenita and Telomere Disorders Panel by next-generation sequencing (NGS)

(*ACD, CTC1, DKC1, NAF1, NHP2, NOP10, PARN, POT1, RTEL1, STN1, TERC, TERF2IP, TERT, TINF2, WRAP53*)

- ☐ Reflex to deletion/duplication of all available genes on panel\*

- ☐ Reflex to deletion/duplication of single gene(s) (specify): \_\_\_\_\_

### ☐ Hemophagocytic Lymphohistiocytosis (HLH) Panel by next-generation sequencing (NGS)

(*AP3B1, AP3D1, CD27, CD70, CTSP1, GATA2, ITK, LYST, MAGT1, NLRC4, PRF1, RAB27A, SH2D1A, SLC7A7, STX11, STXBP2, UNC13D, XIAP (BIRC4)*)

- ☐ Reflex to deletion/duplication of all available genes on panel\*

- ☐ Reflex to deletion/duplication of single gene(s) (specify): \_\_\_\_\_

### Fanconi anemia

- ☐ Fanconi Anemia Panel by next-generation sequencing (NGS)  
(*BRCA1, BRCA2, BRIP1, ERCC4, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, MAD2L2, PALB2, RAD51, RAD51C, RFW3, SLX4, UBE2T, XRCC2*)
- ☐ Reflex to deletion/duplication of all available genes on panel\*
- ☐ Reflex to deletion/duplication of single gene(s) (specify): \_\_\_\_\_

#### ☐ FANCA gene sequencing

- ☐ Reflex to deletion/duplication of FANCA

#### ☐ FANCC gene sequencing

- ☐ Reflex to deletion/duplication of FANCC

- ☐ FANCC c.456+4A>T (IVS4+4 A>T) [common Ashkenazi mutation] only

#### ☐ FANCG gene sequencing

- ☐ Reflex to deletion/duplication of FANCG

### Lymphoproliferative disorders (Including EBV-Related)

#### ☐ SH2D1A gene sequencing

- ☐ Reflex to deletion/duplication of SH2D1A

#### ☐ XIAP (BIRC4) gene sequencing

- ☐ Reflex to deletion/duplication of XIAP (BIRC4)

#### ☐ ITK gene sequencing

- ☐ Reflex to deletion/duplication of ITK

#### ☐ MAGT1 gene sequencing

- ☐ Reflex to deletion/duplication of MAGT1

\*See page 5 for additional deletion/duplication information

## TEST(S) REQUESTED, CONTINUED

### Severe Combined Immunodeficiencies

- ☐ Severe Combined Immunodeficiency panel by next-generation sequencing (NGS)  
(ADA, AK2, ATM, BCL11B, CD247, CD3D, CD3E, CDH17, CHD7, CIITA, CORO1A, DCLRE1C, DOCK8, FOXP1, IL2RG, IL7R, JAK3, LAT, LCK, LIG4, MSN, NHEJ1, ORAI1, PNP, PRKDC, PTPRC, RAC2, RAG1, RAG2, RFX5, RFXANK, RFXAP, RMRP, STAT5B, STIM1, STK4, TAP1, TAP2, TBX1, TTC7A, ZAP70)
- ☐ Add Maternal Engraftment, requires maternal sample of 3 mL blood in EDTA, 2 cytobrushes, or saliva kit.  
Name of mother: \_\_\_\_\_  
DOB (MM/DD/YYYY): \_\_\_\_\_
- ☐ Reflex to deletion/duplication of all available genes on panel<sup>†</sup>
- ☐ Reflex to deletion/duplication of single gene(s)<sup>†</sup> (specify): \_\_\_\_\_
- 
- ☐ IL2RG gene sequencing for X-linked Severe Combined Immunodeficiency
- ☐ Reflex to deletion/duplication of IL2RG

### Severe congenital neutropenia

- ☐ Inherited neutropenia panel by next-generation sequencing (NGS)  
(AK2, AP3B1, CD40LG, CLPB, CSF3R, CXCR2, CXCR4, DNAJC21, EFL1, EIF2AK3, ELANE, G6PC3, GATA1, GATA2, GFI1, HAX1, HYOU1, JAGN1, LAMTOR2, LYST, MRTFA (MKL1), RAB27A, RAC2, RMRP, RUNX1, SBDS, SLC37A4, SMARCD2, SRP54, STK4, TAZ, TCIRG1, TCN2, TP53, USB1, VPS13B, VPS45, WAS, WDR1, WIPF1)
- ☐ Reflex to deletion/duplication of all available genes on panel<sup>†</sup>
- ☐ Reflex to deletion/duplication of single gene(s)<sup>†</sup> (specify): \_\_\_\_\_
- 
- ☐ ELANE gene sequencing
- ☐ Reflex to deletion/duplication of ELANE (ELA2)
- ☐ HAX1 gene sequencing
- ☐ Reflex to deletion/duplication of HAX1
- ☐ WAS gene sequencing (males only)
- ☐ Reflex to deletion/duplication of WAS

### Other Primary Immunodeficiencies

- ☐ FOXP3 gene sequencing for IPEX syndrome
- ☐ Reflex to deletion/duplication of FOXP3
- ☐ WAS gene sequencing for Wiskott-Aldrich syndrome
- ☐ Reflex to deletion/duplication of WAS
- ☐ CD40LG gene sequencing for X-linked hyper IgM immunodeficiency
- ☐ Reflex to deletion/duplication of CD40LG

### Rare Immunodeficiencies

- ☐ CTLA4 gene sequencing
- ☐ Reflex to deletion/duplication of CTLA4
- ☐ GATA2 gene sequencing
- ☐ Reflex to deletion/duplication of GATA2
- ☐ LRBA gene sequencing
- ☐ Reflex to deletion/duplication of LRBA
- ☐ PIK3CD gene sequencing
- ☐ Reflex to deletion/duplication of PIK3CD
- ☐ STAT3 gene sequencing
- ☐ Reflex to deletion/duplication of STAT3
- ☐ Targeted (family specific) variant analysis for \_\_\_\_\_ gene  
If testing was **not** performed at CCHMC, please include proband's report and at least 100ng of proband's DNA to use as a positive control.
- Proband's name \_\_\_\_\_
- Proband's DOB \_\_\_\_\_
- Proband's variant \_\_\_\_\_
- Please call 513-636-4474 to discuss any family-specific variant analysis with genetic counselor prior to shipment.**

<sup>†</sup>See page 5 for additional deletion/duplication information

**Note: Single gene sequencing is available for all genes listed in the next-generation panels.**

## CUSTOM GENE SEQUENCING

Gene(s) to be sequenced (specify): \_\_\_\_\_

**Only genes with clear published functional relationship to rare diseases are accepted.**

Suspected syndrome/ condition: \_\_\_\_\_

**Please choose one of the following:**

- ☐ Full gene(s) sequencing
- ☐ Full gene(s) sequencing with reflex to deletion and duplication analysis, if indicated (**please see list of genes available for del/dup at [www.cincinnatichildrens.org/deldup](http://www.cincinnatichildrens.org/deldup)**)
- ☐ Familial mutation analysis
- Proband's name: \_\_\_\_\_
- Proband's DOB: \_\_\_\_\_
- Proband's mutation: \_\_\_\_\_
- Patient's relation to proband: \_\_\_\_\_
- If testing was **not** performed at CCHMC, please include proband's report and at least 100ng of proband's DNA to use as a positive control.

## DELETION AND DUPLICATION ASSAY

Gene(s) to be analyzed (specify): \_\_\_\_\_

**Please see list of available genes at: [www.cincinnatichildrens.org/deldup](http://www.cincinnatichildrens.org/deldup)**

Suspected syndrome/ condition: \_\_\_\_\_

**Please choose one of the following:**

- ☐ Deletion and duplication analysis of gene(s) specified above
- ☐ Deletion and duplication analysis of gene(s) specified above with reflex to sequencing, if indicated
- ☐ Analysis of gene(s) specified above from previously analyzed deletion and duplication
- ☐ Familial deletion analysis
- Proband's name: \_\_\_\_\_
- Proband's DOB: \_\_\_\_\_
- Proband's mutation: \_\_\_\_\_
- Patient's relation to proband: \_\_\_\_\_
- If testing was **not** performed at CCHMC, please include proband's report and at least 100ng of proband's DNA to use as a positive control.

## IMMUNE DEFICIENCIES, AUTOIMMUNE DISORDERS AND BONE MARROW FAILURE SYNDROMES

### Clinical History is Required for all NGS Panels

#### CLINICAL HISTORY

##### Has patient received a bone marrow transplant?

☐ Yes

☐ No

If yes, date of bone marrow transplant \_\_\_\_\_

Percent engraftment \_\_\_\_\_

##### General

☐ Acute liver failure

☐ Fever(s)

☐ Failure to thrive

☐ (Hepato)splenomegaly

☐ Lethargy

☐ Respiratory insufficiency/failure

☐ Sudden unexplained coma/death

☐ Other; specify \_\_\_\_\_

☐ Age at diagnosis \_\_\_\_\_

##### Head and Neck

☐ Abnormal CT/MRI of brain; specify \_\_\_\_\_

☐ Dysmorphic facies

☐ Enlarged lymph nodes

☐ Microcephaly

☐ Oral leukoplakia

☐ Small lymph nodes and/or tonsils

☐ Thymic hypoplasia

☐ Other; specify \_\_\_\_\_

##### Skin

☐ Alopecia

☐ Eczema

☐ Hypopigmentation/ hyperpigmentation

☐ Rash/dermatitis

☐ Telangiectasia of eyes or skin

☐ Dysplastic nails

☐ Other skin lesions; specify \_\_\_\_\_

##### Hematologic History

☐ Bone marrow failure

☐ Cytopenias (2 of 3 cell lineages)

☐ Leukopenia/neutropenia

☐ Red cell anemia

☐ Thrombocytopenia/small platelets

☐ Other; specify \_\_\_\_\_

##### Oncologic History

☐ Lymphoma; specify type \_\_\_\_\_

☐ Myelodysplasia/AML

☐ Other leukemia; specify type \_\_\_\_\_

☐ Recurrent primary tumors; specify types \_\_\_\_\_

☐ Solid tumor; specify type \_\_\_\_\_

☐ Other; specify \_\_\_\_\_

##### Infectious Disease History

☐ Recurrent, unusual or difficult to treat infections

\_\_\_\_viral \_\_\_\_bacterial \_\_\_\_fungal

☐ Recurrent pneumonia, ear infections or sinusitis

☐ Recurrent deep abscesses of the organs or skin

☐ Multiple courses of antibiotics or IV antibiotics necessary to clear infections

☐ Other; specify \_\_\_\_\_

##### Laboratory findings

☐ Anemia

☐ Decreased telomere length

☐ Neutropenia/leukopenia

☐ Thrombocytopenia

☐ Abnormal ALPS panel

☐ Abnormal mitogen stimulation

☐ Abnormal lymphocyte subsets

☐ Abnormal TREC assay

☐ Abnormal B cell function; specify \_\_\_\_\_

☐ Abnormal T cell function; specify \_\_\_\_\_

☐ Low or absent NK function

☐ Complement group correction (specify) \_\_\_\_\_

☐ Increased chromosome breakage

☐ ↑ ferritin

☐ ↑ soluble IL2Rα

☐ ↑ triglycerides and/or ↓fibrinogens

☐ Abnormal protein assay by flow cytometry; specify \_\_\_\_\_

☐ Other; specify \_\_\_\_\_

##### Congenital abnormalities/malformations/dysmorphic features

(Please specify)

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##### Other Symptoms (Please specify)

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##### Related disease history of other family members (Please specify)

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## ADDITIONAL INFORMATION

### IMMUNOLOGY EXOME PANEL — GENES TESTED

|         |         |         |          |           |          |           |           |         |         |
|---------|---------|---------|----------|-----------|----------|-----------|-----------|---------|---------|
| ACD     | ACP5    | ACTB    | ADA      | ADA2      | ADAM17   | ADAR      | AICDA     | AIRE    | AK2     |
| APIS3   | AP3B1   | AP3D1   | APOL1    | ARPC1B    | ATM      | ATP6AP1   | B2M       | BACH2   | BCL10   |
| BCL11B  | BLM     | BLNK    | BRCA2    | BRIP1     | BTK      | C1QA      | C1QB      | C1QC    | C1R     |
| C1S     | C2      | C3      | C4A      | C4B       | 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     | CDCA7     | CEBPE     | CFB     | CFD     |
| CFH     | CFHR1   | CFHR2   | CFHR3    | CFHR5     | CFI      | CFP       | CFTR      | CHD7    | CIITA   |
| CLCN7   | CLPB    | COG6    | COPA     | CORO1A    | CR2      | CREBBP    | CSF2RA    | CSF2RB  | CSF3R   |
| CTC1    | CTLA4   | CTPS1   | CTSC     | CXCR4     | CYBA     | CYBB      | DCLRE1B   | DCLRE1C | DDX58   |
| DGKE    | DKC1    | DNAJC21 | DNASE1L3 | DNASE2    | DNMT3B   | DOCK2     | DOCK8     | ELANE   | EPG5    |
| ERCC2   | ERCC3   | ERCC4   | ERCC6L2  | EXTL3     | FAAP24   | FADD      | FANCA     | FANCB   | FANCC   |
| FANCD2  | FANCE   | FANCF   | FANCG    | FANCI     | FANCL    | FANCM     | FAS       | FASLG   | FAT4    |
| FCGR3A  | FCN3    | FERMT3  | FOXN1    | FOXP3     | FPR1     | G6PC      | G6PC3     | G6PD    | GATA1   |
| GATA2   | GFI1    | GINS1   | GTF2H5   | HAX1      | HELLS    | HMOX1     | HYOU1     | ICOS    | IFIH1   |
| IFNAR2  | IFNGR1  | IFNGR2  | IGLL1    | IKBKB     | IKZF1    | IL10      | IL10RA    | IL10RB  | IL12B   |
| IL12RB1 | IL17F   | IL17RA  | IL17RC   | IL1RN     | IL21     | IL21R     | IL2RA     | IL2RG   | IL36RN  |
| IL7R    | INO80   | INSR    | IRAK1    | IRAK4     | IRF2BP2  | IRF3      | IRF7      | IRF8    | ISG15   |
| ITCH    | ITGAM   | ITGB2   | ITK      | JAGN1     | JAK1     | JAK3      | KDM6A     | KMT2D   | KRAS    |
| LAMTOR2 | LAT     | LCK     | LIG1     | LIG4      | LPIN2    | LRBA      | LRRC8A    | LYST    | MAGT1   |
| MALT1   | MAN2B1  | MAP3K14 | MASP2    | MBL2      | MCM4     | MEFV      | MKL1      | MPO     | MOGS    |
| MRE11   | MS4A1   | MSH6    | MSN      | MTHFD1    | MVK      | MYD88     | MYH9      | MYO5A   | MYSM1   |
| NBAS    | NBN     | NCF1    | NCF2     | NCF4      | NCSTN    | NFAT5     | NFKB1     | NFKB2   | NFKBIA  |
| NHEJ1   | NHP2    | NLRC4   | NLRP1    | NLRP12    | NLRP3    | NOD2      | NOP10     | NRAS    | NSMCE3  |
| OSTM1   | ORA1    | OTULIN  | PALB2    | PARN      | PCCA     | PCCB      | PEPD      | PGM3    | PIGA    |
| PIK3CD  | PIK3R1  | PLCG2   | PLEKHM1  | PNP       | POLA1    | POLE      | POLE2     | PRF1    | PRKCD   |
| PRKDC   | PSEN1   | PSENEN  | PSMA3    | PSMB4     | PSMB8    | PSTPIP1   | PTEN      | PTPRC   | RAB27A  |
| RAC2    | RAD50   | RAD51C  | RAG1     | RAG2      | RANBP2   | RASGRP1   | RBCK1     | RBM8A   | RELB    |
| RFX5    | RFXANK  | RFXAP   | RHOH     | RNASEH2A  | RNASEH2B | RNASEH2C  | RNF31     | RNF168  | RORC    |
| RPL11   | RPL15   | RPL26   | RPL35A   | RPL36     | RPL5     | RPS10     | RPS15     | RPS15A  | RPS17   |
| RPS19   | RPS24   | RPS26   | RPS27A   | RPS28     | RPS29    | RPS7      | RPSA      | RTKL1   | RUNX1   |
| SAMD9   | SAMD9L  | SAMHD1  | SBDS     | SEMA3E    | SH2D1A   | SH3BP2    | SKIV2L    | SLC29A3 | SLC35A1 |
| SLC35C1 | SLC37A4 | SLC39A4 | SLC46A1  | SLC7A7    | SLX4     | SMARCA1   | SMARCD2   | SNX10   | SP110   |
| SPINK5  | SRP54   | SRP72   | STAT1    | STAT2     | STAT3    | STAT5B    | STIM1     | STK4    | STN1    |
| STX11   | STXBP2  | TAP1    | TAP2     | TAPBP     | TAZ      | TBK1      | TBX1      | TCF3    | TCIRG1  |
| TCN2    | TERC    | TERT    | TFRC     | THBD      | TICAM1   | TINF2     | TIRAP     | TLR3    | TMC6    |
| TMC8    | TMEM173 | TNFAIP3 | TNFRSF1A | TNFRSF11A | TNFSF11  | TNFRSF13B | TNFRSF13C | TNFRSF4 | TNFSF12 |
| TPP2    | TRADD   | TRAF3   | TRAF3IP2 | TREX1     | TRNT1    | TTC37     | TTC7A     | TYK2    | UNC13D  |
| UNC93B1 | UNG     | USB1    | USP18    | VPS13B    | VPS45    | WAS       | WDR1      | WIPF1   | WRAP53  |
| XIAP    | XK      | ZAP70   | ZBTB24   |           |          |           |           |         |         |

\*Targeted deletion and duplication analysis of every gene on this panel except ABCB7, ACD, ACP5, ACTB, ADA2 (CECR1), ADAR, APIS3, AP3D1, APOL1, ARPC1B, ATP6AP1, ATR, B2M, BACH2, BCL10, BCL11B, BRCA1, C4A, C4B, C8G, CARMIL2, CAVIN1, CCBE1, CD46, CD70, CDH17, CFH, CFHR1, CFHR2, CFHR3, CFHR5, CLCN7, CLPB, COPA, CORO1A, CSF2RA, CSF2RB, CTC1, CTPS1, CXCR2, DCLRE1B, DCLRE1C, DDX58, DHFR, DNAJC21, DNASE1L3, DNASE2, DOCK2, EFL1, EIF2AK3, EPG5, EPO, ERCC6L2, ETV6, EXTL3, FAAP24, FANCD2, FAT4, FCGR3A, FERMT3, GINS1, GTF2H5, HELLS, HMOX1, HYOU1, IFIH1, IFNAR2, IGLL1, IKBKB, IL17RC, IL21, INO80, IRAK1, IRF2BP2, IRF3, IRF7, ITGAM, JAGN1, JAK1, KMT2D, LAT, MAD2L2, MAP3K14, MC2R, MRTFA (MKL1), MRE11, MSH6, MSN, MYSM1, NAF1, NBAS, NCF1, NCSTN, NFAT5, NFKB1, NFKB2, NLRC4, NLRP1, NSMCE3, OSTM1, OTULIN, PARN, PEPD, RFWDC3, PGM3, PI4KA, PIGA, PLEKHM1, POLA1, POLE, POLE2, POT1, PRKCD, PROS1, PSEN1, PSENEN, PSMA3, PSMB4, RAD51, RANBP2, RASGRP1, RELB, RNF31, RORC, RPL9, RPL15, RPL18, RPL27, RPL31, RPL36, RPS15, RPS15A, RPS17, RPS27, RPS27A, RPS28, RPS29, RPSA, RUNX1, SAMD9, SAMD9L, SBDS, SEMA3E, SKIV2L, SLC29A3, SLC39A4, SMARCD2, SNX10, SRP54, STAT2, STAT5B, STN1, TCF3, TCIRG1, TCN2, TERF2IP, TFRC, TIRAP, TMEM173, TNFRSF4, TNFSF11, TNFSF12, TP53, TPP2, TRADD, TRAF3IP2, TRNT1, TSR2, UBE, UNC93B1, USP18, WDR1, and XRCC2 is clinically available at an additional charge.