



**Genome Diagnostics – Hereditary Chronic
Kidney Disease**

Toronto General Hospital

Eaton Wing 11-444, 200 Elizabeth Street

Toronto, Ontario M5G 2C4

Phone: (416) 340-4800 x5739/7624

Fax: (416) 340-4473

Hours of Operation (Mon-Fri) 8:30AM-4:30PM

CAP#: 7175217 CLIA#:99D1106115

IQMH# 4204-site 0141

Patient Information or Hospital Stamp Here

Last Name: _____

First Name: _____

Date of Birth (DD/MMM/YYYY): _____

Sex assigned at birth: _____

Health Card #: _____

Hospital #: _____

Instructions:

1. Complete all information as requested
2. Send requisition with specimen to address above
3. Keep specimen at room temperature unless frozen
4. If shipping, send same day or next day delivery
5. Specimen labelling: **Name, DOB, MRN#**

Information for Reporting:

Full Name of Referring physician: _____

Hospital/Address: _____

Phone: _____

Fax: _____

Copy Report To (including fax number) _____

Referring Physician Signature: _____

Specimen Requirements

Peripheral blood (5 mL in EDTA)

Extracted DNA from **BLOOD** only [only accepted from an appropriately accredited laboratory (i.e. ACDx or equivalent)]

Date collected: _____

Conc. _____ Vol. _____

Date collected: _____

Test Indication Please provide any available clinical information and/or complete the Clinical Data Information Sheet.

Diagnosis - symptoms/features of condition in THIS individual; please provide clinical details/eligibility on p2

Known Familial Variant Analysis - Please provide variant details on Pg. 2 of this requisition

****If no family member has been tested at UHN a positive genetic test report of a family member is required.**

Other – Please provide details/justification below

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Hereditary Kidney Disease Panel Testing

(See page 3/4 for panel gene content)

- Amyloidosis (13 genes)
- CAKUT (99 genes)
- Complement Mediated Kidney Disease and Atypical Hemolytic Uremic syndrome (aHUS) (14 genes)
- Cystic Kidney Disease (44 genes)
- Fabry Disease (*GLA* gene)
- Proteinuric Kidney Disease (including Collagenopathies) (80 genes)
- Tubulopathies (including Nephrocalcinosis) (122 genes)
 - Bartter and Gitelman Syndrome (12 genes)
 - Dent Disease (2 genes)
 - Diabetes Insipidus (4 genes)
 - Hypomagnesemia (17 genes)
 - Hypophosphatasia & Inherited Hypophosphatemic Rickets (15 genes)
 - Nephrolithiasis and Nephrocalcinosis (41 genes)
 - Pseudohypoaldosteronism (11 genes)
 - Renal Tubular Acidosis (11 genes)
- Comprehensive Kidney Disease (398 genes)

Familial Variant Analysis

Gene/Variant: _____ Proband Name/UHN MRN _____

(If no family members has been tested at UHN, please include copy of report)

Relationship of this individual to proband: _____

Clinical & Family History

(TESTING MAY BE PUT ON HOLD IF INSUFFICIENT DETAILS PROVIDED)**

PRIMARY Diagnosis: _____

Ethnicity _____

Hereditary Kidney Disease Panel Testing - Sequencing + CNV[†] analysis

[†]copy number variant analysis may result in identification and reporting of copy number variants that involve additional OMIM morbid genes not listed below

Amyloidosis Panel (13 genes): *APOA1, APOA2, APOA4, APOC2, B2M, CST3, FGA, GPNMB, GSN, LYZ, NLRP3, OSMR, TTR*

CAKUT Panel (99 genes): *ACE, ACTG2, AFF3, AGT, AGTR1, ALMS1, ANOS1, BNC2, CCNQ, CDX2, CELSR3, CD151, CENPF, CEP55, CHD7, CHRM3, CHRNA3, COL4A1, CTU2, DDX59, DHCR7, DLG5, DSTYK, DYRK1A, EXOC3L2, EYA1, FAT1, FLCN, FOXC1, FOXP1, FRAS1, FREM1, FREM2, GATA3, GDF6, GFRA1, GLI3, GPC3, GREB1L, GRIP1, HAAO, HNF1B, HOXA13, HPSE2, HS2ST1, HSPA9, ITGA8, JAG1, KAT6b, KDM2B, KCM6A, KIF14, KMT2D, KYNU, LIFR, LRIG2, LRP4, MYOCD, NADSYN1, NEK8, NFIA, NIPBL, NOTCH2, NPHP3, NPNT, NR6A1, NRIP1, OFD1, PAN2, PAX2, PBX1, PLVAP, REN, RET, RMND1, ROBO1, ROBO2, ROR2, SALL1, SALL4, SHROOM4, SLC20A1, SON, STRA6, TBC1D1, TBX18, TBX6, TFAP2A, TMEM260, TOP2B, TRAP1, TXNDC15, WBP11, WLS, WDR72, WNT5A, WT1, ZIC3, ZMYM2*

Complement Mediated Kidney Disease and Atypical Hemolytic Uremic Syndrome (aHUS) Panel (14 genes): *C1GALT1C1, C3, C5, CD46, CFB, CFH, CFHR1, CFHR2, CFHR3, CFHR5, CFI, DGKE, MMACHC, TSEN2*

Cystic Kidney Disease Panel (44 genes): *ALG5, ALG8, ALG9, ANKS6, C2CD3, CEP104, CEP164, CEP83, CLCN5, COL4A1, CYP24A1, DCDC2, DNAJB11, DZIP1L, FLCN, GANAB, GLA, GLIS2, HNF1B, IFT140, INVS, IQCB1, MAPKBP1, MKS1, NEK8, NPHP1, NPHP3, NPHP4, OFD1, PAX2, PKD1, PKD2, PKHD1, PMM2, PRKCSH, SEC63, TMEM67, TSC1, TSC2, TTC21B, TULP3, UMOD, WDR19, XPNPEP3*

Proteinuric Kidney Disease (including Collagenopathies) Panel (80 genes): *ACTN4, ALMS1, AMN, ANLN, APOA1, APOA2, APOA4, APOC2, APOE, APOL1, ARHGDI, CD151, CD2AP, CLCN5, COL4A1, COL4A3, COL4A4, COL4A5, COQ2, COQ6, COQ8B, CRB2, CTNS, CUBN, DAAM2, DGKE, DLC1, FAT1, FN1, GLA, GON7, INF2, ITGA3, ITSN1, ITSN2, KANK2, LAGE3, LAMA5, LAMB2, LCAT, LMX1B, LRP2, MAFB, MAGI2, MTX2, MYH9, MYO1E, NOS1AP, NPHS1, NPHS2, NUP107, NUP133, NUP160, NUP85, NUP93, OCRL, OSGEP, P3H2, PAX2, PBX1, PDSS2, PLCE1, PODXL, PRDM15, PTPRO, REN, SCARB2, SGPL1, SMARCA1, TBC1D8B, TNS2, TP53RK, TPRK, TRIM8, TRPC6, TTC21B, UMOD, WDR73, WT1, YRDC*

Tubulopathies (including Nephrocalcinosis) Panel (122 genes): *AGXT, AIRE, ALPL, AMMECR1, AP2S1, APRT, AQP2, ATP1A1, ATP6VOA4, ATP6V1B1, AVP, AVPR2, BCS1L, BSND, CA2, CACNA1D, CACNA1H, CACNA1S, CASR, CDC73, CDKN1B, CLCN2, CLCN5, CLCNKA, CLCNKB, CLDN10, CLDN16, CLDN19, CNNM2, CPT2, CTNS, CUL3, CYP11B1, CYP11B2, CYP17A1, CYP21A2, CYP24A1, CYP27B1, CYP2R1, DMP1, EHHADH, ENPP1, FAH, FAM111A, FAM20A, FAM20C, FGF23, FOXI1, FXYD2, GALNT3, GATA3, GATM, GCM2, GLA, GNA11, GNAS, GRHPR, HNF1B, HNF4A, HOGA1, HPRT1, HSD11B2, HSD3B2, KCNA1, KCNJ1, KCNJ10, KCNJ16, KCNJ5, KLHL3, LCAT, LRP2, MAGED2, MEN1, MMUT, MOCOS, NDUFAF6, NR3C1, NR3C2, OCRL, PCBD1, PDE3A, PHEX, PTH, PTH1R, REN, RET, RMND1, RRAGD, RRM2B, SARS2, SCN4A, SCNN1A, SCNN1B, SCNN1G, SEC61A1, SLC12A1, SLC12A3, SLC22A12, SLC2A2, SLC2A9, SLC34A1, SLC34A3, SLC3A1, SLC4A1, SLC4A4, SLC5A2, SLC6A1, SLC7A7, SLC7A9, STRADA, STX16, TBCE, TRPM6, TRPM7, UMOD, VDR, VIPAS39, VPS33B, WDR72, WNK1, WNK4, XDH*

Please ensure you are using an updated version of this requisition, available at
https://www.uhn.ca/Labs/services_clinicians#tab2

Hereditary Kidney Disease Panel Testing (cont'd) - Sequencing + CNV⁺ analysis

⁺may result in identification and reporting of copy number variants that involve additional OMIM morbid genes not listed below

Tubulopathies - Subpanels

Bartter and Gitelman Syndrome Panel (12 genes): *BSND, CASR, CLCNKA, CLCNKB, CLDN16, CLDN19, GNA11, HSD11B2, KCNJ1, MAGED2, SLC12A1, SLC12A3*

Dent Disease Panel (2 genes): *CLCN5, OCRL*

Diabetes Insipidus Panel (4 genes): *AQP2, AVP, AVPR2, STRADA*

Hypomagnesemia Panel (17 genes): *ATP1A1, BSND, CASR, CLCNKB, CLDN16, CLDN19, CNNM2, FAM111A, FXSD2, HNF1B, KCNA1, KCNJ10, RRAGD, SARS2, SLC12A3, TRPM6, TRPM7*

Hypophosphatasia and Inherited Hypophosphatemic Rickets Panel (15 genes): *ALPL, CLCN5, CTN2, CYP27B1, CYP2R1, DMP1, ENPP1, FAH, FAM20C, FGF23, OCRL, PHEX, SLC34A1, SLC34A3, VDR*

Nephrolithiasis and Nephrocalcinosis Panel (41 genes): *AGXT, ALPL, AMMECR1, APRT, ATP6VOA4, ATP6V1B1, BSND, CA2, CASR, CLCN5, CLCNKB, CLDN10, CLDN16, CLDN19, CTNS, CYP24A1, FAM20A, GRHPR, HNF4A, HOGA1, HPRT1, KCNJ1, MOCOS, OCRL, PHEX, PTH1R, RRAGD, SLC12A1, SLC22A12, SLC2A9, SLC34A1, SLC34A3, SLC3A1, SLC4A1, SLC7A9, STRAD, VDR, VIPAS39, VPS33B, WDR72, XDH*

Pseudohypoaldosteronism Panel (11 genes): *CUL3, HSD11B2, KCNJ5, KLHL3, NR3C1, NR3C2, SCNN1A, SCNN1B, SCNN1G, WNK1, WNK4*

Renal Tubular Acidosis Panel: (11 genes): *ATP6VOA4, ATP6V1B1, CA2, FOXI1, GATM, HNF4A, RMND1, SLC4A1, SLC4A4, VIPAS39, WDR72*

Comprehensive Kidney Disease Panel (398 genes): All genes currently on the disease-specific panels, plus the following genes associated with ciliopathies, and tubulointerstitial kidney disease: *AHI1, ALG1, ARL13B, ARL6, ARMC9, B9D1, B9D2, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, CC2D2A, CEP290, CEP41, CILK1, CPLANE1, CSPP1, DYNC2H1, DYNC2I1, FAN1, HYLS1, IFT122, IFT172, IFT27, IFT43, IFT74, KATNIP, KIAA0586, KIAA0753, KIF7, LZTFL1, MKKS, MT-TF, MUC1*, NEK1, NPR1, PDIA6, PIBF1, PMPCA, PSKH1, RPGRIP1L, SCLT1, SDCCAG8, TCTN1, TCTN2, TCTN3, TMEM107, TMEM138, TMEM17, TMEM216, TMEM231, TMEM237, IFT54 (TRAF3IP1), TTC8, WDPCP, WDR35, ZNF423*

***the majority of pathogenic MUC1 variants are not detected using current technology; if clinically indicated contact gcConnect for information about additional MUC-1 testing**

FOR ALL HEREDITARY KIDNEY DISEASE TESTS PLEASE ENSURE CLINICAL INFORMATION IS PROVIDED ON PAGE 2

For eligibility please see [OH Genetics Guidance](#)

or contact gcConnect (<https://www.ontariohealth.ca/clinical/genetics/gcconnect>)

Please ensure you are using an updated version of this requisition, available at
https://www.uhn.ca/Labs/services_clinicians#tab2