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Not Tested

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Lawrence M We ss MD
Repor Da e: **May 17,2024**

Access on:
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Tes #: FT TS14824643
Spec men Type: Sa va Swab
Co ec ed: No Prov ded

Access on:
N/A

FINAL RESULTS

Carr er for **ONE** genet c cond t on
Genet c counse ng s
recommended.

TEST PERFORMED

Monash Beacon Expanded Male Carrier Screening Panel v2.1
(363 Gene Pane ; gene sequenc ng w th de et on and dup cat on ana ys s)

Condition and Gene	Inheritance		Partner
Congenital adrenal hyperplasia due to 21-hydroxylase deficiency *CYP21A2*	AR	Poss b e Carr er c 955C>T( )* 2C>T + CYP2 A2 duplication p (Gln3 9*)( )(?)	N/A

INTERPRETATION:

Notes and Recommendations:

- Based on these resu ts, th s nd v dua   s pos t ve for a carr er mutat on n 1 gene. Carr er screen ng for the reproduct ve partner s recommended to accurate y assess the r sk for any autosoma recess ve cond t ons. A negat ve resu t reduces, but does not e m nate, the chance to be a carr er for any cond t on nc uded n th s screen. P ease see the supp ementa tab e for deta s.
- Test ng for copy number changes n the SMN1 gene was performed to screen for the carr er status of Sp na Muscu ar Atrophy. The resu ts for th s nd v dua are w th n the norma range for non-carr ers. See L m tat ons sect on for more nformat on.
- Th s carr er screen ng test does not screen for a   poss b e genet c cond t ons, nor for a   poss b e mutat ons n every gene tested. Th s report does not nc ude var ants of uncerta n s gn f cance; on y var ants c ass f ed as pathogen c or   ke y pathogen c at the t me of test ng, and cons dered re evant for reproduct ve carr er screen ng, are reported. P ease see the gene spec f c notes for deta s. P ease note that the c ass f cat on of var ants can change over t me.
- Pat ents may w sh to d scuss any carr er resu ts w th b ood re at ves, as there s an ncreased chance that they are a so carr ers. These resu ts shou d be nterpreted n the context of th s nd v dua 's c n ca f nd ngs, b ochem ca prof e, and fam y h story.
- X- nked genes are not rout ne y ana yzed for ma e carr er screen ng tests. Gene spec f c notes and   m tat ons may be present. See be ow.
- Genet c counse ng s recommended. Contact your phys c an about the ava ab e opt ons for genet c counse ng.

CONGENITAL ADRENAL HYPERPLASIA DUE TO 21-HYDROXYLASE DEFICIENCY

Patient		Partner
Result	Poss b e Carr er	N/A
Variant Details	***CYP21A2*** (NM_000500.9) c.955C>T(;)*12C>T + CYP21A2 dup cat on p.(G n319*)(;)(?)	N/A

What is Congenital adrenal hyperplasia due to 21-hydroxylase deficiency?

Congen ta adrena hyperp as a (CAH) due to 21-hydroxy ase def c ency s an nher ted d sorder that affects the adrena g ands and hormone product on. Approx mate y 75 percent of nd v dua s w th c ass c 21-hydroxy ase def c ency have the sa t-wast ng type, whereby the body excretes too much sa t n ur ne. Affected nfants present w th poor feed ng, we ght oss, dehydrat on, and vom t ng, a of wh ch can be fe-threaten ng. Fema es w th th s cond t on typ ca y have amb guous gen ta a, wh e ma es usua y have norma gen ta a, but w th sma testes. Ind v dua s w th the s mp e v r z ng form and the non-c ass c form of the d sease do not exper ence sa t oss. Ma es and fema es w th e ther the c ass c or non-c ass c forms of 21-hydroxy ase def c ency tend to have an ear y growth spurt, but the r f na adu t he ght s usua y shorter than others n the r fam y, and affected nd v dua s may have reduced fert ty. Add t ona y, nd v dua s may have excess ve body ha r growth, ha r oss, and rregu ar menstruat on. Some nd v dua s (ma e or fema e) w th the non-c ass c form of the d sease may have m d, non- fe-threaten ng symptoms, wh e others may never deve op symptoms of the d sorder at a .

What is my risk of having an affected child?

CAH due to 21-hydroxy ase def c ency s nher ted n an autosoma recess ve manner. If the pat ent and the partner are both carr ers, the r sk for an affected ch d s 1 n 4 (25%).

What kind of medical management is available?

Treatment cons sts of ear y n t at on of hormone rep acement therapy and/or surgery for fema es. Prognos s s good for pat ents w th appropr ate med ca management and psycho og ca support.

What mutation was detected?

The heterozygous var ants c.955C>T (p.G n319*) and a who e gene dup cat on of CYP21A2 were detected n th s samp e. In add t on, the ben gn po ymorph sm c.*12C>T was a so detected. The phase of these var ants s unknown but cou d be determ ned through parenta test ng.

The nonsense var ant, p.G n319*, ntroduces a premature stop codon and s expected to resu t n the oss of funct on of the prote n product of the CYP21A2 gene, e ther as the resu t of prote n truncat on or of nonsense-med ated mRNA decay. Th s var ant, a so reported as Q318*, s a c ass c 21-hydroxy ase-def c ent congen ta adrena hyperp as a mutat on and has been reported n mu t p e affected nd v dua s (PubMed: 3267225, 12220458, 12915679). The var ant, p.G n319*, and the po ymorph sm c.*12C>T are known to frequent y occur n a dup cated copy of the CYP21A2 gene coex st ng w th a norma copy of CYP21A2 on the same chromosome. Th s hap otype was dent f ed n approx mate y 2% of the genera popu at on and n ~80% of carr ers of p.G n319*, and such a conf gurat on may represent a ben gn a e e (PubMed: 28401898, 19773403). Nonethe ess, there s a poss b ty that p.G n319* occurs on a chromosome w th on y a s ng e copy of CYP21A2, n wh ch case t resu ts n a pathogen c a e e. If mu t p e cop es of CYP21A2 are present, we cannot be certa n f th s p.G n319* var ant occurs on a chromosome w th one (.e. pathogen c state) or two (.e. ben gn state) cop es of CYP21A2. Wh e th s comb nat on of var ants may represent a ben gn a e e, the aboratory c ass f es the var ant p.G n319* as ke y pathogen c.

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GENES TESTED:

Monash Beacon Expanded Male Carrier Screening Panel v2.1 - 363 Genes
Th s ana ys s was run us ng the Monash Beacon Expanded Ma e Carr er Screen ng Pane v2.1 gene st. 363 genes were tested w th 99.5% of targets sequenced at >20x coverage. For more gene-spec f c nformat on and ass stance w th res dua r sk ca cu at on, see the SUPPLEMENTAL TABLE.

ABCA12 ABCA3 ABCA4 ABCB11 ABCC8 ACAD9 ACADM ACADVL ACAT1 ACOX1 ACSF3 ADA ADAMTS2 ADGRG1 ADK AGA AGL AGPS AGXT AHI1 AIPL1 ALDH3A2 ALDOB ALG6 ALMS1 ALPL AMT AQP2 ARG1 ARL13B ARSA ARSB ASL ASNS ASPA ASS1 ATM ATP6V1B1 ATP7B BBS1 BBS10 BBS12 BBS2 BCKDHA BCKDHB BCS1L BLM BSND CAPN3 CASQ2 CBS CC2D2A CCDC103 CCDC39 CCDC88C CDH23 CEP290 CFTR CHRNE CHRNG CHST6 CIITA CLN3 CLN5 CLN6 CLN8 CLRN1 CNGB3 COL27A1 COL4A3 COL4A4 COL7A1 COX15 CPS1 CPT1A CPT2 CRB1 CRYL1 CTNS CTSA CTSC CTSD CTSK CYBA CYP11A1 CYP11B1 CYP11B2 CYP17A1 CYP1B1 CYP21A2 CYP27A1 DBT DCLRE1C DDX11 DHCR7 DHDDS DLD DNAH5 DNAI1 DNAI2 DUOX2 DUOXA2 DYNC2H1 DYSF EIF2AK3 EIF2B5 ELP1 ERCC2 ERCC5 ERCC6 ERCC8 ESCO2 ETFA ETFB ETFDH ETHE1 EVC EVC2 EXOSC3 F2 F5 FAH FAM126A FAM161A FANCA FANCC FANCG FH FKRP FKTN FOXRED1 FTCD FUCA1 G6PC GAA GALC GALNS GALT GAMT GBA GBE1 GCDH GDAP1 GDF5 GFM1 GJB2 GJB6 GLB1 GLDC GLE1 GNE GNPTAB GNPTG GNS GSS GUCY2D GUSB HADHA HADHB HAX1 HBA1 HBA2 HBB HEXA HEXB HGSNAT HJV HLCS HMGCL HOGA1 HPS1 HPS3 HPS4 HSD17B4 HSD3B2 HYLS1 IDUA IVD IYD JAK3 KCNJ11 LAMA2 LAMA3 LAMB3 LAMC2 LCA5 LDLRAP1 LHX3 LIFR LIPA LMBRD1 LOXHD1 LPL LRP2 LRPPRC LYST MAN2B1 MANBA MCOLN1 MCPH1 MED17 MESP2 MFSD8 MKS1 MLC1 MLYCD MMAA MMAB MMACHC MMADHC MPI MPL MPV17 MTHFR MTMR2 MTRR MTTP MUT MVK MYO7A NAGA NAGLU NAGS NBN NDRG1 NDUFAF2 NDUFAF5 NDUFS4 NDUFS6 NDUFS7 NDUFV1 NEB NEU1 NPC1 NPC2 NPHP1 NPHS1 NPHS2 NTRK1 OAT OCA2 OPA3 OTOF P3H1 PAH PANK2 PC PCCA PCCB PCDH15 PCNT PDHB PEX1 PEX10 PEX12 PEX2 PEX26 PEX6 PEX7 PFKM PHGDH PHYH PKHD1 PLA2G6 PLOD1 PMM2 POLG POLR1C POMGNT1 POMT1 POMT2 POR PPT1 PRF1 PROP1 PSAP PTS PUS1 QDPR RAB23 RAG1 RAG2 RAPSN RARS2 RAX RDH12 RMRP RNASEH2B RPE65 RPGRIP1L RTEL1 SACS SAMD9 SAMHD1 SCO2 SEPSECS SERPINA1 SGCA SGCB SGCD SGCG SGSH SH3TC2 SLC12A6 SLC17A5 SLC19A3 SLC1A4 SLC22A5 SLC25A13 SLC25A15 SLC26A2 SLC26A3 SLC35A3 SLC37A4 SLC39A4 SLC45A2 SLC46A1 SLC5A5 SLC7A7 SMARCAL1 SMN1 SMPD1 SPG11 SPINK5 STAR SUMF1 SURF1 TCIRG1 TCTN2 TECPR2 TF TG TGM1 TH TMEM216 TPO TPP1 TRDN TRIM32 TRMU TSEN54 TSFM TSHB TTC37 TTPA TYMP TYR TYRP1 UGT1A1 USH1C USH1G USH2A VPS13A VPS13B VPS45 VPS53 VRK1 VSX2 WHRN WRN XPA XPC ZFYVE26

METHODS:

Genom c DNA was so ated from the subm tted spec men nd cated above (f ce u ar mater a was subm tted). DNA was barcoded, and enr ched for the cod ng exons of targeted genes us ng hybr d capture techno ogy. Prepared DNA brar es were then sequenced us ng a Next Generat on Sequenc ng techno ogy. Fo ow ng a gnment to the human genome reference sequence (assemb y GRCh37), var ants were detected n reg ons of at east 10x coverage. For th s spec men, 99.54% and 99.49% of cod ng reg ons and sp c ng junct ons of genes sted had been sequenced w th coverage of at east 10x and 20x, respect ve y, by NGS or by Sanger sequenc ng. The rema n ng reg ons d d not have 10x coverage, and were not eva uated. Var ants were nterpreted manua y us ng ocus spec f c databases, terature searches, and other mo ecu ar b o og ca pr nc p es. To m n m ze fa se pos t ve resu ts, any var ants that do not meet nterna qua ty standards are conf rmed by Sanger sequenc ng. Var ants c ass f ed as pathogen c, ke y pathogen c, or r sk a e e wh ch are ocated n the cod ng reg ons and nearby ntron c reg ons (+/- 20bp) of the genes sted above are reported. Var ants outs de these nterva s may be reported but are typ ca y not guaranteed. When a s ng e pathogen c or ke y pathogen c var ant s dent f ed n a c n ca y re evant gene w th autosoma recess ve nher tance, the aboratory w attempt to ensure 100% coverage of cod ng sequences e ther through NGS or Sanger sequenc ng techno og es ("f - n"). A genes sted were eva uated for arge de et ons and/or dup cat ons. However, s ng e exon de et ons or dup cat ons w not be detected n th s assay, nor w copy number a terat ons n reg ons of genes w th s gn f cant pseudogenes. Putat ve de et ons or dup cat ons are ana yzed us ng Fu gent Germ ne propr etary p pe ne for th s spec men. B o nformat cs: The Fu gent Germ ne v2019.2 p pe ne was used to ana yze th s spec men.

LIMITATIONS:

General Limitations

These test resu ts and var ant nterpretat on are based on the proper dent f cat on of the subm tted spec men, accuracy of any stated fam a re at onsh ps, and use of the correct human reference sequences at the quer ed oc . In very rare nstances, errors may resu t due to m x-up or co-m ng ng of spec mens. Pos t ve resu ts do not mp y that there are no other contr butors, genet c or otherw se, to future pregnanc es, and negat ve resu ts do not ru e out the genet c r sk to a pregnancy. Off c a gene names change over t me. Fu gent uses the most up to date gene names based on HUGO Gene Nomenc ature Comm ttee (https://www.genenames.org) recommendat ons. If the gene name on report does not match that of ordered gene, p ease contact the aboratory and deta s can be prov ded. Resu t nterpretat on s based on the ava ab e c n ca and fam y h story nformat on for th s nd v dua , co ected pub shed nformat on, and A amut annotat on ava ab e at the t me of report ng. Th s assay s not des gned or va dated for the detect on of ow- eve mosa c sm or somat c mutat ons. Th s assay w not detect certa n types of genom c aberrat ons such as trans ocat ons, nvers ons, or repeat expans ons other than spec f ed genes. DNA a terat ons n regu atory

reg ons or deep ntron c reg ons (greater than 20bp from an exon) may not be detected by th s test. Un ess otherw se nd cated, no add t ona assays have been performed to eva uate genet c changes n th s spec men. There are techn ca m tat ons on the ab ty of DNA sequenc ng to detect sma nsert ons and de et ons. Our aboratory uses a sens t ve detect on a gor thm, however these types of a terat ons are not detected as re ab y as s ng e nuc eot de var ants. Rare y, due to systemat c chem ca , computat ona , or human error, DNA var ants may be m ssed. A though next generat on sequenc ng techno og es and our b o nformat cs ana ys s s gn f cant y reduce the confound ng contr but on of pseudogene sequences or other h gh y-homo ogous sequences, somet mes these may st nterfere w th the techn ca ab ty of the assay to dent fy pathogen c a terat ons n both sequenc ng and de et on/dup cat on ana yses. De et on/dup cat on ana ys s can dent fy a terat ons of genom c reg ons wh ch nc ude one who e gene (bucca swab spec mens and who e b ood spec mens) and are two or more cont guous exons n s ze (who e b ood spec mens on y); s ng e exon de et ons or dup cat ons may occas ona y be dent f ed, but are not rout ne y detected by th s test. When nove DNA dup cat ons are dent f ed, t s not poss b e to d scern the genom c ocat on or or entat on of the dup cated segment, hence the effect of the dup cat on cannot be pred cted. Where de et ons are detected, t s not a ways poss b e to determ ne whether the pred cted product w rema n n-frame or not. Un ess otherw se nd cated, de et on/dup cat on ana ys s has not been performed n reg ons that have been sequenced by Sanger.

Gene Specific Notes and Limitations

CEP290: Copy number ana ys s for exons 8-13 and exons 39-42 may have reduced sens v y n he CEP290 gene. Conf rma on of hese exons are m ed o nd v dua s w h a pos ve persona h s ory of CEP290-re a ed cond ons and/or nd v dua s carry ng a pa hogen c/ ke y pa hogen c sequence var an . *CFTR:* Ana ys s of he n ron 8 po ymorph c reg on (e.g. IVS8-5T a e e) s on y performed f he p.Arg117 s (R117) mu a on s de ec ed. S ng e exon de e on/dup ca on ana ys s s m ed o de e ons of prev ous y repor ed exons: 1, 2, 3, 11, 19, 20, 21. CFTR var an s pr mar y assoc a ed w h CFTR-re a ed so a ed congen a b a era absence of he vas deferens and CFTR-re a ed pancrea s are no nc uded n h s ana ys s. CFTR var an s w h nsuff c en ev dence of be ng cys c f bros s mu a ons w no be repor ed e her. *CRYL1:* As mu a ons n he CRYL1 gene are no known o be assoc a ed w h any c n ca cond on, sequence var an s n h s gene are no ana yzed. owever, o ncrease copy number de ec on sens v y for arge de e ons nc ud ng h s gene and a ne ghbor ng gene on he pane (GJB6, a so known as connex n 30), h s gene was eva ua ed for copy number var a on. *CYP11B1:* The curren es ng me hod s no ab e o re ab y de ec cer a n pa hogen c var an s n h s gene due o he n erference by h gh y homo ogous reg ons. Th s ana ys s s no des gned o de ec or ru e-ou copy-neu ra ch mer c CYP11B1/CYP11B2 gene. *CYP11B2:* The curren es ng me hod s no ab e o re ab y de ec cer a n pa hogen c var an s n h s gene due o he n erference by h gh y homo ogous reg ons. Th s ana ys s s no des gned o de ec or ru e-ou copy-neu ra ch mer c CYP11B1/CYP11B2 gene. *CYP21A2:* S gn f can pseudogene n erference and/or rec proca exchanges be ween he CYP21A2 gene and s pseudogene, CYP21A1P, have been known o occur and may mpac resu s. As such, he re evance of var an s repor ed n h s gene mus be n erpre ed c n ca y n he con ex of he c n ca f nd ngs, b ochem ca prof e, and fam y h s ory of each pa en . CYP21A2 var an s pr mar y assoc a ed w h non-c ass c congen a adrena hyperp as a (CA) are no nc uded n h s ana ys s (PubMed: 23359698). The var an s assoc a ed w h non-c ass c d sease, nc ud ng bu no m ed o c.188A>T (p. s63Leu), c.844G>T (p.Va 282Leu), c.1174G>A (p.A a392Thr), and c.1360C>T (p.Pro454Ser) w no be repor ed. LR-PCR s no rou ne y ordered for NM 000500.9:c.955C>T (p.G n319Ter). Ind v dua s w h c.955C>T (p.G n319Ter) w be repor ed as a Poss b e Carr er nd ca ng ha he prec se na ure of he var an has no been de erm ned by LR-PCR and ha he var an may occur n he CYP21A2 w d- ype gene or n he CYP21A1P pseudogene. The conf rma on es s recommended f he second reproduc ve par ner s es ed pos ve for var an s assoc a ed w h c ass c CA . *DDX11:* Due o he n erference by h gh y homo ogous reg ons, our curren es ng me hod has ess sens v y o de ec var an s n he DDX11 gene. *DUOX2:* The curren es ng me hod s no ab e o re ab y de ec var an s n exons 6-8 of he DUOX2 gene (NM 014080.5) due o s gn f can n erference by he h gh y homo ogous gene, DUOX1. *F2:* The common r sk a e e NM 000506.5:c.*97G>A s no nc uded n h s ana ys s. *F5:* The common Fac or 5 "Le den" a e e s no yp ca y repor ed as h s var an s assoc a ed w h ow d sease pene rance. *GALT:* In genera , he D2 "Duar e" a e e s no repor ed f de ec ed, bu can be repor ed upon reques . Wh e h s a e e can cause pos ve newborn screen ng resu s, s no known o cause c n ca symp oms n any s a e (PubMed: 25473725, 30593450). *GBA:* The curren es ng me hod may no be ab e o re ab y de ec cer a n pa hogen c var an s n he GBA gene due o homo ogous recomb na on be ween he pseudogene and he func ona gene. *HBA1:* S gn f can n erference from h gh y homo ogous reg ons n exons 1-2 of he BA1 gene has been recogn zed o occur, po en a y mped ng he assay's echn ca capab y o de ec pa hogen c a era ons dur ng sequenc ng ana yses. *HBA2:* S gn f can n erference from h gh y homo ogous reg ons n exons 1-2 of he BA2 gene has been recogn zed o occur, po en a y mped ng he assay's echn ca capab y o de ec pa hogen c a era ons dur ng sequenc ng ana yses. *HSD17B4:* Copy number ana ys s for exons 4-6 may have reduced sens v y n he SD17B4 gene. Conf rma on of hese exons are m ed o nd v dua s w h a pos ve persona h s ory of D-b func ona pro e n def c ency and Perrau syndrome and/or nd v dua s carry ng a pa hogen c/ ke y pa hogen c sequence var an . *LMBRD1:* Copy number ana ys s for exons 9-12 may have reduced sens v y n he LMBRD1 gene. Conf rma on of hese exons are m ed o nd v dua s w h a pos ve persona h s ory of comb ned me hy ma on c ac dur a and homocys nur a and/or nd v dua s carry ng a pa hogen c/ ke y pa hogen c sequence var an . *MTHFR:* As recommended by ACMG, he wo common po ymorph sms n he MT FR gene - c.1286A>C (p.G u429A a, a so known as c.1298A>C) and c.665C>T (p.A a222Va , a so known as c.677C>T) - are no repor ed n h s es due o ack of suff c en c n ca u y o mer es ng (PubMed: 23288205). *NEB:* Th s gene con a ns a 32-kb r p ca e reg on (exons 82-105) wh ch

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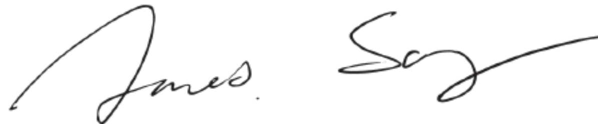
s no amenab e o sequenc ng and de e on/dup ca on ana ys s. ***NPHS2:*** If de ec ed, he var an NM 014625.3:c.686G>A (p.Arg229G n) w no be repor ed as h s var an s no s gn f can y assoc a ed w h d sease when homozygous or n he compound he erozygous s a e w h var an s n exons 1-6 of NP S2. ***SERPINA1:*** If de ec ed he var an NM 000295.5:c.863A>T (p.G u288Va) w no be repor ed as h s var an s assoc a ed w h ow d sease pene rance and s no assoc a ed w h severe ear y onse d sease. ***SMN1:*** The curren es ng me hod de ec s sequenc ng var an s n exon 7 and copy number var a ons n exons 7-8 of he SMN1 gene (NM 022874.2). Sequenc ng and de e on/dup ca on ana ys s are no performed on any o her reg on n h s gene. Abou 5%-8% of he popu a on have wo cop es of SMN1 on a s ng e chromosome and a de e on on he o her chromosome, known as a [2+0] conf gura on (PubMed: 20301526). The curren es ng me hod canno d rec y de ec carr ers w h a [2+0] SMN1 conf gura on, bu can de ec nkage be ween he s en carr er a e e and cer a n popu a on-spec f c s ng e nuc eo de changes. As a resu , a nega ve resu for carr er es ng grea y reduces bu does no e m na e he chance ha a person s a carr er. On y abnorma resu s w be repor ed. ***TRDN:*** Due o h gh GC con en of cer a n exons (nc ud ng exons 4-5), copy number ana ys s may have reduced sens v y for par a gene de e ons/dup ca ons of *TRDN*. Conf rma on of par a gene de e ons/dup ca ons are m ed o nd v dua s w h a pos ve persona h s ory of card ac arrhy hm a and/or nd v dua s carry ng a pa hogen c/ ke y pa hogen c sequence var an . ***TYR:*** Due o he n erference by h gh y homo ogous reg ons, our curren es ng me hod has ess sens v y o de ec var an s n exons 4-5 of he TYR gene (NM 000372.5). ***UGT1A1:*** Common var an s n he *UGT1A1 gene* (popu a on a e e frequency >5%) are yp ca y no repor ed as hey do no cause a Mende an cond on. ***VPS45:*** LoF s no a known d sease mechan sm ***WRN:*** Due o he n erference by h gh y homo ogous reg ons w h n he WRN gene, our curren es ng me hod has ess sens v y o de ec var an s n exons 10-11 of WRN (NM 000553.6).

SIGNATURE:



Jianbo Song, Ph.D., ABMGG, CGMB, CCS, FACMG on **5/17/2024**
Labora ory D rec or Fu gen

DISCLAIMER:

Th s test was deve oped and ts performance character st cs determ ned by **Fulgent Therapeutics LLC**. It has not been c eared or approved by the FDA. The aboratory s regu ated under CLIA as qua f ed to perform h gh-comp ex ty test ng. Th s test s used for c n ca purposes. It shou d not be regarded as nvest gat ona or for research. S nce genet c var at on, as we as systemat c and techn ca factors, can affect the accuracy of test ng, the resu ts of test ng shou d a ways be nterpreted n the context of c n ca and fam a data. For ass stance w th nterpretat on of these resu ts, hea thcare profess ona s may contact us d rect y at **(626) 350-0537** or **info@fulgentgenetics.com**. It s recommended that pat ents rece ve appropr ate genet c counse ng to exp a n the mp cat ons of the test resu t, nc ud ng ts res dua r sks, uncerta nt es and reproduct ve or med ca opt ons.

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To view the supplemental table describing the carrier frequencies, detection rates, and residual risks associated with the genes on this test please visit the following link:
Beacon Expanded Carrier Screening Supplemental Table