Now Available...

Comprehensive services to identify the cause of infections from nail and skin specimens

Now available, a comprehensive menu of pathology testing plus molecular testing for dermatology and podiatry practices. Utilizing advanced testing, our board-certified pathologists provide rapid, reliable test results to aid in diagnosing or screening for dermatologic conditions.

- Skin and Soft Tissue Infections by Molecular Analysis
- Hereditary Melanoma Cancer Panel for the identification of disease-causing genetic variants
- Pharmacogenomic Testing to predict a patient's genetic influences on the dosing, toxicities, and efficacy of specific drugs

Advantages:

- One vial, multiple pathogens
- DNA amplification via PCR technology
- Simple & convenient specimen collection
- High precision robotic accuracy
- High diagnostic sensitivity & specificity
- Specimen viability up to 5 days after collection
- Test additions available up to 30 days after collection
- No refrigeration required before or after collection
- **Highly skilled Dermatopathologist**

4path Pathology Services

Gene & Transcript	Variant	Inheritance	Disorder or Phenotype	Criteria	Classification
BRCA2 NM_000059.3	c.1841_1842dup Asn615fs	Autosomal Recessive / Homozygous	Hereditary cancer-predisposing syndrome	PM2, PVS1, PP5	Likely Pathogenic
Location	**Allele State**	**Allelic Read Depths**			
Exon 10	Het.	Ref(T): , Alt(TT): , VAF: NaN%			
Genomic Position		**Variant Frequency**			
NM_000059.3:		Not identified in large population studies			

VARIANT INTERPRETATION:
The frameshift duplication NM_000059.4(BRCA2):c.1841_1842dupTT (p.Asn615Leufs*30) has been reported to ClinVar as Likely Pathogenic with a status of (1 stars) criteria provided, single submitter (Variation ID 633102 as of 2021-04-01). The p.Asn615Leufs*30 variant is novel (not in any individuals) in gnomAD. The p.Asn615Leufs*30 variant is novel (not in any individuals) in 1kG. This variant is predicted to cause loss of normal protein function through protein truncation caused a frameshift mutation. The frame shifted sequence continues 30 residues until a stop codon is reached. This variant is a frameshift variant which occurs in an exon of BRCA2 upstream of where nonsense mediated decay is predicted to occur. There are 2764 downstream pathogenic loss of function variants, with the furthest variant being 2805 residues downstream of this variant. Thi
the gene BRCA2, which is intolerant of Loss of Func
NP_000050.3:p.M1Kfs*3419 and 3342 others. For t

Gene & Transcript	Variant
RB1 NM_000321.3	c.74C>T p.Pro25Leu
Location	**Allele State**
Exon 1	Het.
	Genomic Position
	NM_000321.3:

VARIANT INTERPRETATION: The missense varian
benign variant, to our knowledge. The p.Pro25Leu v
There is a moderate physicochemical difference bet
missense variants Z-Score of 2.67. The gene RB1 c
in this gene. For these reasons, this variant has bee

METHODOLOGY

The individual's DNA was extracted and fragmented,
amplification and sequencing. Reads from the seque
Sentieon DNAseq (vs. 2018.08.07). The variants wer
guidelines for interpretation of sequence variants. Th
Project Consortium's publication of 2,500 genomes,
evidence on conservation and functional impact.
The variant results are reported using numbering and
codon number are based on the gene transcript: **BA**
CDKN2A(NM_000077.4), **MITF**(NM_001354604.2),

In addition to the gene sequencing assay, a MLPA ar
***BRCA1** and **BRCA2** genes*, was completed.

VARIANT ASSESSMENT PROCESS

The following databases and *in silico* algorithms are
ClinVar, OMIM, dbSNP, NCIB RefSeq Genes, ExAC
Splice Predictor. Analysis was reported using the HG
reported transcript matches that used most frequentl

LIMITATIONS

It should be noted that this test is performed on a lim
affecting gene regions not examined in the assay. Int
report only includes variants that meet a level of evid
the NGS testing technology, including triplet repeat e
More evidence for disease association of genes and
interpreted with updated software and annotations pe
This test was developed and its performance charact
analytical performance of the test. It is not been revie
not necessary.

REFERENCES

Richards, Sue, et al. "Standards and guidelines for th
Genetics and Genomics and the Association for Mole
Exome Aggregation Consortium et al. "Analysis of Pr
The 1000 Genomes Project Consortium. "A Global R

Mail:	Yes	USP
	None	Yes

MEDICAL DIAGNOSTIC LABORATORIES

2439 KUSER ROAD, HAMILTON, NJ 08690-3303
TL: 609 890 1000, FX: 609 890 1050, TF: 877 269 0090
www.mdlab.com

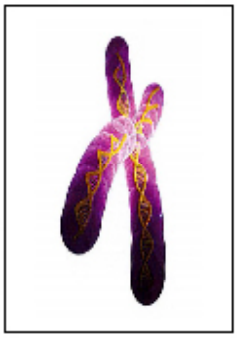
MDL#: 9649360 Final Test Results Physician Copy

Patient Information: SSN: XXX-XX-0001 DOB: 12/1974 (Age: 44)
DOE, JANE
123 MAIN ROAD
ANYTOWN, NJ 08888
Home: (888) 888-8888 Patient ID:

Ordering Physician/Lab: NPI: 1234567890
DOE WOMANS GROUP
JOHN DOE, MD
555 SMITH STREET
ANYTOWN, NJ 55555
Tel: (856) 555-555
Fax: (856) 555-5552
Results Faxed To:
IN

Genetic Counselor Information
Genetic counseling was refused
IN
Tel: 681-322-1902
Fax: 85-322-190

Specimen Type:	Saliva
Date Collected:	4/26/2021
Date Received:	4/27/2021
Date Reported:	4/30/2021



BRCAcare + 2605 Melanoma Panel

RESULT: Positive for Deleterious Mutation

AFFECTED GENES

BAP1 (0)	BRCA1 (0)	BRCA2 (1)	CDK4 (0)	CDKN2A (0)	MITF (0)	POT1 (0)	PTEN (0)	RB1 (1)	TP53 (0)

VARIANTS RELEVANT TO INDICATION FOR TESTING

Gene & Transcript	Variant	Allele State	Location	Disorder or Phenotype	Inheritance	Classification
BRCA2 NM_000059.3	c.1841_1842dup Asn615fs	Het.	Exon 10	Hereditary cancer-predisposing syndrome	Autosomal Recessive / Homozygous	Likely Pathogenic
RB1 NM_000321.3	c.74C>T p.Pro25Leu	Het.	Exon 1	Unspecified	Autosomal Dominant / Heterozygous	Uncertain Significance

RECOMMENDATIONS

The interpretation of these results should be done in the context of a patient's medical record and family history. Please note that interpretation and classification of the variants reported here may change over time. Please see a genetic counselor for services regarding the implications of these results in the context of understanding the implications of incidental findings, family planning and the informing of family members of potentially shared genetic outcomes.

APPROACH

Sequencing of the coding regions and flanking non-coding regions of select genes was performed using Next Generation Sequencing and the data was analyzed to identify both previously classified and novel variants in targeted genes. The select gene panel including targeted genes with previous implications of association with breast cancer, ovarian cancer, and/or Lynch syndrome were covered with a minimum read depth of 20X. A multiplex ligation-dependent probe amplification (MLPA) analysis which detects deletions and/or duplications involving one or more exons of ***BRCA1*** and ***BRCA2*** genes was completed.

DETAILED VARIANT INFORMATION (VARIANTS RELEVANT TO INDICATION FOR TESTING)

Mail:	Yes	USPS	Fax:	Yes	Manual
	None	Yes		None	No

Medical Director: Jing-Jing Yang, M.D

1/30/2021
Final