McLastname, First N
Patient ID: **123456789**
Specimen ID: **119-990-4002-0**

DOB: **06/01/1947**
Age: **72**
Sex: **Male**

Account Number: **01818355**
Ordering Physician: **David Johnson**

labcorp

Corrected Report - Prenatal Spinal Muscular Atrophy (SMA)

 # genes, including add-on gene(s) for F2 (1 variant), F5 (1 variant), F8, F9, G6PD, GP1BA, GP9, HFE, SERPINA1, TFR2

Specimen Type: **Saliva**
Fetus ID: **Twin A**

Ethnicity: **Ashkenazi Jewish, Caucasian**
Indication: **Carrier Test**

Genetic Counselor: **Michael Mull**

Date Collected: **04/29/2019 1142 Local**
Date Received: **04/29/2019 1620 ET**
Date Reported: **04/30/2019 1045 ET**
Date Entered: **06/03/2019 1045 ET**

<Corrected Comments>

Summary: Positive

Positive Results

DISORDER (GENE)	RESULTS	INTERPRETATIONS
Alpha-1 antitrypsin deficiency (SERPINA1) NM_XXXXXXXXX	<< Result >>	<< Interpretation >> Risk: <<RISK>>
Gaucher disease (GBA) NM_001005741	<< Result >>	<< Interpretation >> Risk: <<RISK>>

Negative Results

DISORDER (GENE)	RESULTS	INTERPRETATIONS
Cystic fibrosis (CFTR) NM_000492	<< Result >>	<< Interpretation >> Risk: <<RISK>>
Fragile X syndrome (FMR1) NM_002024	<< Result >>	<< Interpretation >> Risk: <<RISK>>
Spinal muscular atrophy (SMN1) NM_000344	<< Result >>	<< Interpretation >> Risk: <<RISK>>
ALL OTHER DISORDERS	<< Result >>	This result reduces, but does not eliminate the risk to be a carrier. **Risk**: The individual is NOT at an increased risk for having a pregnancy that is affected with one of the disorders covered by this test. For partner's gene-specific risks, visit www.integratedgenetics.com.

< General comments section - need to be able to enter text manually or populate with MCC output from RightReport for prenatal report types >
Comparison of maternal and fetal DNA markers indicates that maternal cell contamination is unlikely to have interfered with reported fetal result (maternal specimen # XXXXXXXXXX)

Recommendations

Genetic counseling is recommended to discuss the potential clinical and/or reproductive implications of positive results, as well as recommendations for testing family members and, when applicable, this individual's partner. Genetic counseling services are available. To access Integrated Genetics Genetic Counselors please visit www.integratedgenetics.com/genetic-counseling or call (855) GC-CALLS (855-422-2557).

Additional Clinical Information

Fragile X syndrome: Fragile X syndrome is an X-linked disorder of intellectual disability with variable severity. Expansions of CGG repeat sequences in the FMR1 gene account for 99% of mutations causing fragile X syndrome. The risk of expansion from a premutation allele of 55-90 repeats to a full mutation in offspring, when transmitted by a carrier female, is reduced with increasing number of AGG interruptions in the CGG repeat sequence (Yrigollen, PMID:22498846; Nolin, PMID:25210937). Greater than 99% of males and approximately 50% of females with the full mutation are intellectually disabled. Other signs and symptoms may include delayed speech and language skills, autism, hyperactivity, developmental delay, increased susceptibility to seizures, macroorchidism in males, a long, narrow face with prominent ears, and joint laxity. Individuals with a premutation do not have fragile X syndrome, but may have an increased risk for fragile X-related disorders. Females may have fragile X-associated primary ovarian insufficiency (FXPOI), which can cause infertility or early menopause. Most males with a premutation and some females are at risk for fragile X-associated tremor and ataxia syndrome (FXTAS), which can affect balance and is associated with tremor and memory problems in older individuals. Treatment is supportive and focuses on educational and behavioral support and management of symptoms. (Santoro, PMID:22017584).

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Corrected Report - Prenatal Spinal Muscular Atrophy (SMA)

 # genes, including add-on gene(s) for F2 (1 variant), F5 (1 variant), F8, F9, G6PD, GP1BA, GP9, HFE, SERPINA1, TFR2

Comments

This interpretation is based on the clinical information provided and the current understanding of the molecular genetics of the disorder tested. References and additional information about the disorders tested are available at www.integratedgenetics.com.

Methods/Limitations

Fragile X syndrome: Repeat-primed PCR is used to detect the number of CGG repeats on each allele of the FMR1 gene. The reportable range is 5-200 repeats. Alleles with expansions above 200 repeats are reported as >200. In females, excluding prenatal specimens, alleles between 55 and 90 repeats are assessed by a PCR assay to determine the number and position of AGG interruptions within the CGG repeats. Interpretation of repeat expansion results is based on the following ranges: Negative: < 45 repeats; intermediate: 45-54 repeats; premutation: 55-200 repeats; full mutation: >200 repeats. The analytical sensitivity of this assay for the detection of expanded alleles in the FMR1 gene is estimated to be >99%. Repeat numbers are typically 1 for alleles containing up to 60 repeats, 3 for alleles containing 61-119 repeats, and 10 for alleles with >119 repeats. Low levels of mosaicism (<5%) and FMR1 variants unrelated to trinucleotide expansion are not detected by this assay.

Information Table

CF detection rates for prenatal testing		
Disorder (gene) Reference sequence	**Population**	**Detection rate**
Cystic fibrosis (CTFR) NM_000492	African American	81%
	Ashkenazi Jewish	97%
	Asian American	55%
	Caucasian	93%
	Hispanic	78%
	Mixed or other ethnic background	For counseling purposes, consider using the ethnic background with the most conservative estimates.

References

1.Eaton, T. V., & Akers, M. D. (2007). Whistleblowing and good governance. CPA Journal, 77(6), 66–71.
2. American Nurses Association. (2015). Nursing: Scope and standards of practice (3rd ed.).

Disordered Tested

3-Methylcrotonyl-CoA carboxylase deficiency (2 genes). Autosomal recessive: MCCC1, MCCC2.
3M syndrome (3 genes). Autosomal recessive: CCDC8, CUL7, OBSL1.
Abetalipoproteinemia (1 gene). Autosomal recessive: MTTP.
Acute infantile liver failure (3 genes). Autosomal recessive: LARS, NBAS, TRMU.
Adenosine deaminase deficiency (1 gene). Autosomal recessive: ADA.
Adrenoleukodystrophy, X-linked (1 gene). X-Linked: ABCD1. Males are not tested for X-linked disorders.
Agammaglobulinemia, X-linked (1 gene). X-Linked: BTK. Males are not tested for X-linked disorders.
Aicardi-Goutières syndrome (4 genes). Autosomal recessive: RNASEH2A, RNASEH2B, RNASEH2C, SAMHD1.
Allan-Herndon-Dudley syndrome (1 gene). X-Linked: SLC16A2. Males are not tested for X-linked disorders.
Alpha-mannosidosis (1 gene). Autosomal recessive: MAN2B1.
Alpha-thalassemia X-linked intellectual disability syndrome (1 gene). X-Linked: ATRX. Males are not tested for X-linked disorders.
Alport syndrome, X-linked (1 gene). X-Linked: COL4A5. Males are not tested for X-linked disorders.
Alström syndrome (1 gene). Autosomal recessive: ALMS1.

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 # genes, including add-on gene(s) for F2 (1 variant), F5 (1 variant), F8, F9, G6PD, GP1BA, GP9, HFE, SERPINA1, TFR2

Patient Details
McLastname, FirstName N
401 Holly Street NW, Atlanta, GA 30318
Phone: **336-436-0515**
Patient ID: **1234**
Alternate Patient ID: **ALTP2318975**

Physician Details
David Johnson
Remedy Hospital
2550 McMillan Parkway Burlington, NC 27215
Account Number: **01818355**
Physician ID:
NPI:

Specimen Details
Specimen ID: **119-990-4002-0**
Control ID:
Alternate Control Number: **ALTC8975456**

Performing Labs

Component Type	Performed at	Laboratory Director
Technical component, processing	Esoterix Genetic Laboratories, LLC, 3400 Computer Drive, Westborough, MA 01581	Bernice Allitto, PhD, FACMG
Technical component, analysis	Laboratory Corporation of America, 1912 TW Alexander Drive, RTP, NC 27709-0150	Anjen Chenn, MD, PhD
Professional component	Esoterix Genetic Laboratories, LLC, 3400 Computer Drive, Westborough, MA 01581	Bernice Allitto, PhD, FACMG

For inquires, the physician may contact the lab at (800) 255-7357

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