

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

Ethnicity: Ashkenazi Jewish, Caucasian

Genetic Counselor: Michael Mull

Fetus ID: Twin A

Indication: Carrier Test

Date Collected: 04/29/2019

Date Received: 04/29/2019

Date Reported: 04/30/2019

Summary: Negative

DISORDER (GENE)	RESULTS	INTERPRETATIONS
Spinal muscular atrophy (SMN1) NM_000344	<< Result >>	<< Interpretation >>

< 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 # XXXXXXXXXXXX)

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](http://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).

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](http://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.

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Summary: Positive

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Patient Details	Physician Details	Specimen ID 119-990-4002-0
First McLastname	David Johnson	Control ID
401 Holly Street NW, Atlanta, GA 30318	Remedy Hospital	Date Collected 06/01/2019 1142 Local
	2550 McMillan Parkway Burlington, NC 27215	Date Received 06/02/2019 1620 ET
Phone 336-436-0515	Phone 336-436-0515	Date Entered 06/03/2019 1045 ET
Date of Birth 06/01/47	Account Number 01818355	Date Reported 06/04/2019 1045 ET
Age 72	Physician ID	Rte: 00
Sex Male	NPI	
Patient ID 1234	Referring Physician George Smith	