McLastname, First N
Patient ID: **1234**
Specimen ID: **119-990-4002-0**

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

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

labcorp

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

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

Genetic Counselor: **Michael Mull**

Date Collected: **04/29/2019**
Date Received: **04/29/2019**
Date Reported: **04/30/2019**

Tests Ordered: **Test Name**

SUMMARY: <NEGATIVE or POSITIVE>

POSITIVE RESULTS or NEGATIVE RESULTS

DISORDER (*GENE*)	RESULTS	INTERPRETATION
Spinal muscular atrophy (*SMN1*) NM_000344	<<RESULT>>	<<INTERPRETATION>>

<General comments section - need to be able to enter text manually or populate with MCC info 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 the 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

Spinal muscular atrophy: Spinal muscular atrophy (SMA) is an autosomal recessive neurodegenerative disorder with variable age at onset and severity, characterized by progressive degeneration of the lower motor neurons in the spinal cord and brain stem, leading to muscle weakness, and in its most common form, respiratory failure by age two. Complications of SMA may include poor weight gain, sleep difficulties, pneumonia, scoliosis, and joint deformities. In severely affected individuals, abnormal fetal ultrasound findings may include congenital joint contractures, polyhydramnios, and decreased fetal movement (Korinthenberg, PMID:9307259). Treatment is supportive. Targeted therapies may be available for some individuals. Approximately 94% of affected individuals have 0 copies of the *SMN1* gene; in these individuals an increase in the number of copies of the *SMN2* gene correlates with reduced disease severity (Feldkotter, PMID:11791208). Individuals with one copy of the *SMN1* gene are predicted to be carriers of SMA; those with two or more copies have a reduced carrier risk. For individuals with two copies of the *SMN1* gene, the presence or absence of the variant c.*3+80T>G correlates with an increased or decreased risk, respectively, of being a silent carrier (2+0) (Luo, PMID 23788250; Feng, PMID 28125085).

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

Spinal muscular atrophy: The copy number of SMN1 exon 7 is assessed relative to internal standard reference genes by quantitative polymerase chain reaction (qPCR). A mathematical algorithm calculates 0, 1, 2 and 3 copies with statistical confidence. When no copies of *SMN1* are detected, the primer and probe binding sites are sequenced to rule out variants that could interfere with copy number analysis and *SMN2* copy number is assessed by digital droplet PCR analysis relative to an internal standard reference gene. For carrier screening, when two copies of *SMN1* are detected, allelic discrimination qPCR targeting c.*3+80T>G in *SMN1* is performed.

Electronically released under the direction of <Director Name, Certification>

CF detection rates for prenatal testing		
Disorder *(Gene)* Reference sequence	**Population**	**Detection rate**
Cystic fibrosis (*CFTR*) 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.