NTD | Eurofins Clinical Diagnostics

Patient Information — Final Report

PATIENT NAME: [redacted]
DATE OF BIRTH: [redacted]
CROSS REFERENCE #: [redacted]
REFERRING DIAGNOSIS: [redacted]
PATIENT ID: [redacted]
LABORATORY #: [redacted]

TYPE OF SPECIMEN: [redacted]
DATE COLLECTED: [redacted]
DATE RECEIVED: [redacted]
FINAL REPORT: [redacted]

REFERRING PHYSICIAN: [redacted]
ADDRESS: [redacted]
TELEPHONE: [redacted]
FAX NUMBER: [redacted]
REFERRING PRACTICE: [redacted]
TELEPHONE: [redacted]
FAX NUMBER: [redacted]

Spinal Muscular Atrophy: Carrier Screen

Results: POSITIVE. One copy of the common *SMN1* gene deletion associated with spinal muscular atrophy (SMA) was detected.

Interpretation

A sample from this individual was submitted to our laboratory for carrier testing for spinal muscular atrophy (SMA). SMA is an autosomal recessive condition involving progressive muscle weakness due to degeneration of the lower motor neurons in the spinal cord and brain stem. Onset and severity can vary and classification of SMA subtype is commonly made based by age of onset and maximum function achieved. SMA is caused by pathogenic variants in the *SMN1* gene. Approximately 95-98% of SMA is caused by a homozygous deletion of the *SMN1* gene, approximately 2% of which occur *de novo*. The other 2-5% is caused by one copy of an *SMN1* gene deletion and one copy of an *SMN1* small pathogenic variant. This analysis is for the common *SMN1* deletion only.

One copy of the common *SMN1* gene deletion associated with spinal muscular atrophy (SMA) was detected. This result indicates that this individual is a carrier of SMA. This condition is inherited in an autosomal recessive manner which means both parents have to be carriers to have a 1 in 4 (25%) risk to have an affected child. Genetic counseling is recommended.

SMA carrier testing is available at EGL Genetics for this individual's partner and family members at risk for carrying the *SMN1* gene deletion identified in this individual. For more information, please visit eglgenetics.com or call (470) 378-2200 to contact a laboratory genetic counselor or consult with a laboratory director.

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Notes: Direct detection of *SMN1* deletions is highly accurate. Possible diagnostic errors include sample mix-ups, genotyping errors, rare genetic variants which interfere with analysis, and other sources.

Methodology: Isolated DNA from this individual was evaluated for *SMN1* gene deletions using multiplex ligation polymerase chain reaction amplification (MLPA) of exons 7 and 8 according to the SALSA protocol available from MRC Holland. The gene dosage ratio was calculated relative to the average of 16 reference loci. Two copies of *SMN1* most likely indicate normal (not affected) status and one copy of *SMN1* most likely indicates carrier status. Other pathogenic variants in *SMN1*, such as small pathogenic variants and fusion *SMN* genes, will not be detected by this assay.

Reference: Hendrickson et al. J Med Genet. 2009 Sep;46(9):641-4.

Medical Consultant — Laboratory Director, Molecular Genetics

This case has been reviewed and electronically signed by a Laboratory Director.