TARP SYNDROME

RBM10 GENE SEQUENCING

TARP syndrome (OMIM 311900) is an X-linked disorder caused by mutations in the RBM10 gene (OMIM 300080). The hallmark symptoms of TARP syndrome include talipes equinovarus, atrial septal defect, Robin sequence, and persistence of the left superior vena cava. Other common symptoms are low-set ears, cryptorchidism, and pulmonary hypoplasia. The majority of affected males die in early infancy, although there is one report (Gripp 2011) of an affected male who has survived into early childhood. Additional features were noted in this child, including sensorineural hearing loss, optic atrophy and cortical visual impairment, motor retardation, and severe intellectual disability. To date, female carriers have not been shown to exhibit any symptoms related to the disease.

Testing:
Testing is performed by sequencing the entire coding region of the RBM10 gene. This will detect point mutations, small deletions, and small insertions. It will not detect a partial or whole gene deletion or duplication. To date, only nonsense and frameshift mutations with single nucleotide insertions or deletions have been reported.

RBM10 is the only gene known to be associated with TARP syndrome. This test will only detect mutations in RBM10.

Turn-around time: 10-14 business days

CPT codes and cost: 83891 x 1  83900 x 1  83901 x 2  83904 x 39  83912 x 1  $1500

BILLING: We do not bill third party payers (insurance companies) for samples received from external sources. The person or institution (Clinical Lab; Send-out Lab; Physician Office) sending the sample is responsible for full payment of the invoices within 30 days of receipt of the invoice. If the patient is on Medical assistance, please contact the lab prior to sample submission. Direct patient billing will only be accepted when a valid credit card form is received with the patient sample.

Online resources:
OMIM entry for RBM10: http://omim.org/entry/300080
OMIM entry for TARP syndrome: http://omim.org/entry/311900

References:


Facsimile Verification Form

Name of Facility receiving Fax: ____________________________________________
Name of Physician/Lab receiving Fax: ________________________________________
Street Address: ____________________________________________________________
City __________________________ State: _____
Fax Number: ____________________________________________
(to which lab results and/or patient information may be sent)
Phone Number: ____________________________________________

By signing this Facsimile Verification Form, I validate the accuracy of the above information and assume responsibility for assuring that the Fax machine is in a location which will maintain confidentiality of all reports transmitted by the Molecular Diagnostics Laboratory of the Alfred I. duPont Hospital for Children, to the above fax number.

Authorized Contact Person: ________________________________________________
Signature: __________________________ Date: ______________
Title: __________________________________________________________

In our continuing efforts to maintain patient confidentiality, the Molecular Diagnostics Laboratory of the Alfred I. duPont Hospital for Children requests you to verify the fax number only once from your medical practice or institution and to assure that all faxes regarding patient information are received in a secure location in accordance with HIPAA regulations.

Please complete this Facsimile Verification Form and fax back to 302.651.6795.
If you have any questions regarding this form please contact Susan Kirwin, Assistant Director of The Molecular Diagnostics Laboratory, at 302.651.6777.

This is a confidential document that is being sent from a fax machine in a secure location. This fax is covered by the Electronic Communications Privacy Act 18 U.S.C. 2510-521. The information contained in this fax is considered privileged, is otherwise confidential and is intended only for the use of the individual or entity named above. Dissemination, distribution, or copying of this communication is strictly prohibited. If you have received this communication in error please notify us immediately by telephone, and return the original message to us at the above address via the U.S. Postal Service.
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<th>Credit Card Billing Information</th>
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For Direct Patient Billing
Prepayment for the testing services is required prior to beginning our testing. Please complete this form and include this paperwork with the shipment of the patient sample.

Billing questions can be addressed to: Denise Axsmith
Senior Budget/Financial Analyst
Nemours/A.I. duPont Hospital for Children
daxsmith@nemours.org
Phone: 302.651.6802
Fax: 302.651.6881