ESCOPBAR SYNDROME

CHRNNG GENE SEQUENCING

Escobar syndrome (OMIM 265000) is an autosomal recessive disorder caused by changes in the CHRNNG gene (OMIM 100730). Escobar syndrome is one of a group of disorders known as multiple pterygium syndromes. These disorders are characterized by arthrogryposis (joint contractures) and pterygia (webbing) of the neck, elbows, and/or knees. Other features include respiratory distress, scoliosis, dysmorphic facial features, short stature, cleft palate, and cryptorchidism in males. Reduced fetal movement is frequently reported.

Lethal multiple pterygium syndrome (OMIM 253290) is a more severe form of this disorder and can also be caused by mutations in the CHRNNG gene. Arthrogryposis and multiple pterygia are severe and most individuals are stillborn or die in the neonatal period, primarily due to pulmonary hypoplasia. Intrauterine growth retardation, cystic hygroma, and hydrops may be present. In addition to the findings described in Escobar syndrome, findings in lethal multiple pterygium syndrome include cardiac hypoplasia and generalized amyoplasia.

**Testing:** Testing is performed by sequencing the entire coding region and the surrounding intronic regions of the CHRNNG gene. This will detect point mutations, small deletions, and small insertions. It will not detect a partial or whole gene deletion or duplication.

CHRNNG is the only gene known to be associated with Escobar syndrome. Lethal multiple pterygium syndrome has been associated with CHRNNG mutations, as well as mutations in the related genes, CHRNA1 and CHRNND.

**Turn-around time:** 10-14 days

**CPT codes and cost:**
83891 (x1)  83900 (x1)  83901 (x1)  83898 (x1)  83904 (x16)  83912 (x1)  $700

**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.* Direct patient billing will only be accepted when a valid credit card form is received with the patient sample. If the patient is on Medical assistance, please contact the lab prior to sample submission.

**Online resources:**
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 Diagnostic 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 Diagnostic 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 Diagnostic Laboratory, at 302.651.6777.

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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  
[duxsmith@nemours.org](mailto:duxsmith@nemours.org)  
Phone: 302.651.6802  
Fax: 302.651.6881