MICROCEPHALIC OSTEODYSPLASTIC PRIMORDIAL DWARFISM TYPE II

*PCNT* MUTATION ANALYSIS

Microcephalic osteodysplastic primordial dwarfism type II (MOPD II; OMIM 210720) is an autosomal recessive disorder caused by mutations in *PCNT* (OMIM 605925). MOPD II is characterized by progressive intrauterine growth retardation (IUGR) which can be recognized as early as 13 weeks gestation. At term, infants typically weigh less than 3 lbs and are less than 16 inches in length. Most children are born slightly premature at 35 weeks with an average birth weight of 2 lbs 3 oz.

Relative microcephaly is not typically present at birth, but head size becomes disproportionately small as children grow. The trunk, chest, and spine are proportionately small, with disproportionately small forearms. Radiologic features may be subtle in the newborn period. As children age, the bones appear thin and delicate with progressive widening of the metaphyses. Other characteristics include a prominent nose and eyes, small teeth with enamel defects, fine sparse hair, and pigmentary dysplasia of the skin. About 20% of individuals with MOPD II have abnormalities in the cerebral vascular system which can predispose to stroke.

**Testing:** Testing for MOPDII is performed by sequencing all exons and surrounding intronic regions. This test will detect point mutations, small deletions, and small insertions. It will not detect a partial or whole gene deletion or duplication.

*PCNT* is the only gene known to be associated with MOPD II. A negative test does not rule out a diagnosis of MOPD II since a mutation could be found in a region not sequenced or in another gene.

**Turnaround time:** 4 weeks or less

**CPT codes and cost:**
89891(x1)  83898(x5)  83900(x1)  83901(x 7)  83904(x 100)  83912 (x1)  $ 2200

**BILLING:** *We do not bill third party payers (insurance companies)* for samples received from external sources. [The person or institution (e.g., 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 be accepted *only* when a valid credit card form is received with the patient sample.

**Online Resources:**
Potential Foundation (support organization) - [http://www.potentialsfoundation.org/](http://www.potentialsfoundation.org/)

**References:**

Facsimile Verification Form

Name of Facility receiving Fax: ____________________________________________________________
Name of Physician/Lab receiving Fax: _____________________________________________________

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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.

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telephone, and return the original message to us at the above address via the U.S. Postal Service.
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

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