



Children's Hospital Boston  
Department of Neurology



HARVARD MEDICAL SCHOOL  
Department of Neurology

Children's Hospital Boston  
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RE:	DATE:
Rachel Vontungeln	7/21/2009
TOTAL NUMBER OF PAGES INCLUDING COVER:	2
NOTES/COMMENTS:	

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CLIA #22D0883928  
New York State #CQP4866

REPORT OF DNA-BASED TEST RESULTS

Test: Juvenile Neuronal Ceroid-lipofuscinosis (JNCL, Batten disease) DNA analysis

Patient Name: Rachel Vontungeln  
Patient ID number: DOB 5/18/00  
Sample submitted: DNA  
Date of specimen collection:  
Date of specimen receipt: 7/13/09  
Accession #: 09-803  
Date of Report: 7/17/09

Referring Physician/Counselor:  
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RESULTS:

Genomic DNA from this individual was analyzed by polymerase chain reaction (PCR) amplification of an internal fragment of the Batten disease gene (*CLN3*) in both directions. DNA fragments were sized by agarose gel electrophoresis. Only a 1.0 kb deleted fragment was identified.

INTERPRETATION:

DNA testing shows this individual to be homozygous for the Batten's associated deletion.

As reported recently<sup>1</sup>, a 1.02kb genomic deletion encompassing intronic sequence and 2 coding exons of *CLN3* has been identified in approximately 73% of alleles of patients or obligate carriers of Batten disease (JNCL). Further DNA testing for carrier and/or affected status is possible in this family and should be considered if appropriate. Genetic counseling is recommended.

Testing of at-risk family members is available. Genetic counseling is recommended.

A support group is available for patients and families with NCL disorders. If interested, please contact Mr. Lance Johnston at 800-448-4570.

Winnie W. Xin, Ph.D.  
Co-Director

Katherine B. Sims, M.D.  
Director

References: <sup>1</sup>The International Batten Disease Consortium (1995) Isolation of a novel gene underlying Batten disease, *CLN3*. Cell 82:949-957.



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