SCHOOL OF MEDICINE
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National Prion Disease Pathology Surveillance Center
Department of Pathology
2085 Adelbert Road, Room 418
Cleveland, OH 44106-4907
216-368-0587 main
216-368-2546-fax
216-368-4090-confidential

Fax

To:	Dr. Jeanette Larson	From:	NPDPSC
Phone:		Phone:	216.368.0587
Fax:	828-213-9914	Fax:	216.368.4090
Pages:	2 including cover	Date:	
Re:	NPDPSC # 2014-1593	CC:	Dr. Proia: 919-684-2625 Dr. Stephens: 828-253-4830 Dr. Ryan: 866-285-9740 Dr. Schneider: 828-213-9914 NC DOH: 919-733-9555

☐ Urgent ☐ For Review ☐ Please Comment ☐ Please Reply ☐ Please Recycle

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The National Prion Disease Pathology Surveillance Center

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Dr. Bernardino Ghetti, Co-Director
Dr. Jiri Safar, Co-Director
Dr. Wenquan Zou, Associate Director
Dr. Clive Hamlin, Director, Clinical Laboratory
Dr. Shulin Zhang, Genetic Analyses
Dr. Mark Cohen, Histopathology
Andrea Webb, Center Manager

Advisory Committee:
Dr. Barbara Crain
Dr. Stephen DeArmond
Dr. Daniel Peri

Dr. Jeanette Larson
Mission Health
Neurology
509 Biltmore Avenue
Asheville, North Carolina 28801

10/6/2014

Dear Dr. Larson,

We have completed the analyses on the autopsy tissue that you sent us from Laura Yavelow (your #14-174-508701/ AO-14-206; our #O2014-1593; DOB 9/13/1950). We have characterized the abnormal prion protein type by Western blot and carried out the histopathological and immunohistochemical examinations. The sequencing of the prion protein (PrP) gene is based on the report issued by Dr. Shulin Zhang of the Center for Human Genetics of University Hospitals of Cleveland. The results of these tests performed on the case that you submitted confirm the diagnosis of prion disease with the characteristics of sporadic Creutzfeldt-Jakob disease (sCJD)MM1 according to the classification of sporadic prion disease proposed by Parchi et al. (Annals of Neurology 46: 224-233, 1999). The PrP gene sequencing rules out the presence of a pathogenic mutation in the coding region of the PrP gene. Therefore, the prion disease in the case you submitted is not familial according to the current criteria for familial prion diseases (Kong et al. Prion Biology and Diseases 2 ed. Prusiner, S. Editor., 2004. 673-775).

Polymerase Chain Reaction (PCR) amplification followed by sequence analysis of a DNA sample from this individual was used. This method does not constitute a definitive diagnostic test or a precise test for prion associated diseases. Possible sources of diagnostic error include sample mix-up and genotyping errors. Genotyping errors can result from trace contamination of PCR reactions, from rare genetic variants which interfere with analysis, and from other sources. Individuals being studied should understand that rare diagnostic errors would occur for these reasons.
These assays are for investigational purposes and should be used in conjunction with other clinical, pathological, and laboratory findings.
On behalf of the National Prion Disease Pathology Surveillance Center, I thank you for submitting this case to us for review. We look forward to receiving future cases of possible prion disease.

Thank you for referring to us this interesting case.

Sincerely yours,

Pierluigi Gambetti M.D.

PG/ats

cc: Alan Proia, Duke University Medical Center
cc: Alexander Schneider, Mission Health
cc: Lisa Ryan, Mission Health
cc: Michael Stephens, The Family Health Centers
cc: Marilyn Goss Haskell, North Carolina Department of Health and Human Services

Institute of Pathology
Case Western Reserve University
2085 Adelbert Road
Cleveland, Ohio 44106-4907

Telephone 216-368-0587
Fax 216-368-4090
E-Mail cjdsurv@case.edu
Website www.cjdsurveillance.com