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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
216-368-4080-confidential

Fax

To: Dr. Jeanette Larson

From: NPDPS

Phone:

Phone: 216.368.0587

Fax: 828-213-9914

Fax: 216.368.4090

Pages: 2 including cover

Date:

Re: NPDPS # 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

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The National Prion Disease Pathology Surveillance Center

Sponsored by the American Association of Neuropathologists (AANP)

Office:

Dr. Pierluigi Gambetti, Director
 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 Perl

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

10/6/2014

Dear Dr. Larson,

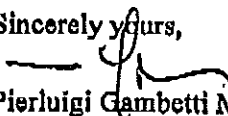
We have completed the analyses on the autopsy tissue that you sent us from Laura Yavelow (your #14-174-508701/AO-14-206; our #O2014-1593; DOB 9/13/1950). We have characterized the abnormal prion protein type by Western blot and carried out the histopathological and immunohistochemical examinations. The 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

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

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