ADDENDUM

CC2D2A, Encoding A Coiled-Coil and C2 Domain Protein, Causes Autosomal-Recessive Mental Retardation with Retinitis Pigmentosa

Abdul Noor, Christian Windpassinger, Megha Patel, Beata Stachowiak, Anna Mikhailov, Matloob Azam, Muhammad Irfan, Andrew D. Paterson, Muhammad Lutufullah, Dan Doherty, John B. Vincent,* and Muhammad Ayub

(American Journal of Human Genetics *82*, 1011–1018; April 2008)

In our recent report,[1] brain magnetic resonance imaging (MRI) data for the oldest (male) and the youngest (female) affected members of the Mianwali family were used to initially diagnose the patients with nonsyndromic mental retardation. Although the molar tooth sign (MTS) was observed in the MRI of the female, suggesting a Joubert syndrome-related disorder (JSRD, MIM 213300), a diagnosis of JSRD was overruled because of the lack of any other obvious JSRD features. We have since reviewed the MRI images with D. Doherty (University of Washington), and mid-hindbrain features typical of JSRD, including the MTS, are present in both of these individuals (Figure 4E in Gorden et al., this issue[2]). This information, together with data from the JSRD families described by Gorden et al., would suggest that the mutation in *CC2DA* in our Mianwali family is most likely a cause of a form of JSRD.

References

1. Noor, A., Windpassinger, C., Patel, M., Stachowiak, B., Mikhailov, A., Azam, M., Irfan, M., Siddiqui, Z.K., Naeem, F., Paterson, A.D., et al. (2008). CC2D2A, encoding a coiled-coil and C2 domain protein, causes autosomal-recessive mental retardation with retinitis pigmentosa. Am. J. Hum. Genet. *82*, 1011–1018.
2. Gorden, N.T., Arts, H.H., Parisi, M.A., van Beersum, S.E.C., Hikida, A., Letteboer, S.J.F., Eckert, M., Coene, K.L.M., Knutzen, D., Mans, D.A., et al. (2008). *CC2D2A* is mutated in Joubert syndrome and interacts with the ciliopathy-associated, basal body protein CEP290. Am. J. Hum. Genet. *83*, this issue, 559–571.

*Correspondence: john_vincent@camh.net
DOI 10.1016/j.ajhg.2008.10.005.