Molecular Genetics and Metabolism Reports 2 (2015) 80

Contents lists available at ScienceDirect

Molecular Genetics and Metabolism Reports

journal homepage: http://www.journals.elsevier.com/molecular-genetics-and-metabolism-reports/

Letter to the Editor

Stormorken syndrome or York platelet syndrome: A clinician's dilemma

Keywords:
Stormorken syndrome
York platelet syndrome
Miosis
Tubular aggregate myopathy

To the Editor:

We read the paper recently published by Markello et al. in the latest issue [1]. Analysis of our observations [2] together with a review of other cases [3,4] suggests that patients with the c.910C>T mutation in *STIM1* gene (p.R304W) have the same heterogeneous disease and that Stormorken syndrome (StS) and York platelet syndrome (YPS) are probably two different names for the same clinical condition. The confusion could be due to the strategy used to study platelets. Indeed, in patients affected with YPS platelets have been extensively studied by electron microscopy, not in the patients with StS. It should be noted that among the many biological symptoms of StS, thrombocytopenia and thrombocytopathia have been first described as major and constant defects [2–5].

Patients of families C and D carry the typical c.910C>T mutation also observed in StS. But the occurrence of miosis and the spleen anomalies have not been reported. Conversely, patients of families A and B bearing a different mutation (c.343A>T) located in EF-hand domain, do exhibit miosis and spleen anomalies. One YPS patient has been diagnosed for tubular aggregate myopathy (TAM). Muscle complaints and histopathological results found in YPS patients are identical to those observed in isolated TAM and StS patients. This suggests that other patients with TAM due to mutation of EF-hands could have other symptoms belonging to StS spectrum [6,7]. The rimmed vacuoles observed in another YPS patient resemble tubular aggregates and require further investigations.

Based on the similar mutations and identical clinical phenotype, we suggest renaming these clinical conditions as the Stormorken–York platelet syndrome. Identifying the underlying genetic cause including heterogeneity in modifiers still remains a challenge.

Acknowledgment

This study was supported by the Direction of the Clinical Research and Innovation of the CHU d' Amiens.

References

[1] T. Markello, D. Chen, J.Y. Kwan, I. Horkayne-Szakaly, A. Morrison, O. Simakova, I. Maric, J. Lozier, A.R. Cullinane, T. Kilo, L. Meister, K. Pakzad, W. Bone, S. Chainani, E. Lee, A. Links, C. Boerkoel, R. Fischer, C. Toro, J.G. White, W.A. Gahl, M. Gunay-Aygun, York platelet syndrome is a CRAC channelopathy due to gain-of-function mutations in STIM1, Mol. Genet. Metab. (Dec 24 2014) (pii: S1096-7192(14)00700-8).
[2] G. Morin, N.O. Bruechle, A.R. Singh, C. Knopp, G. Jedraszak, M. Elbracht, D. Brémond-Gignac, K. Hartmann, H. Sevestre, P. Deutz, D. Hérent, P. Nürnberg, B. Roméo, K. Konrad, M. Mathieu-Dramard, J. Oldenburg, E. Bourges-Petit, Y. Shen, K. Zerres, H. Ouadid-Ahidouch, J. Rochette, Gain-of-function mutation in STIM1 (p.R304W) is associated with Stormorken syndrome, Hum. Mutat. 35 (10) (Oct 2014) 1221–1232.
[3] V. Nesin, G. Wiley, M. Kousi, E.C. Ong, T. Lehmann, D.J. Nicholl, M. Suri, N. Shahrizaila, N. Katsanis, P.M. Gaffney, K.J. Wierenga, L. Tsiokas, Activating mutations in STIM1 and ORAI1 cause overlapping syndromes of tubular myopathy and congenital miosis, Proc. Natl. Acad. Sci. U. S. A. 111 (11) (Mar 18 2014) 4197–4202.
[4] D. Misceo, A. Holmgren, W.E. Louch, P.A. Holme, M. Mizobuchi, R.J. Morales, A.M. De Paula, A. Stray-Pedersen, R. Lyle, B. Dalhus, G. Christensen, H. Stormorken, G.E. Tjønnfjord, E. Frengen, A dominant STIM1 mutation causes Stormorken syndrome, Hum. Mutat. 35 (5) (May 2014) 556–564.
[5] H. Stormorken, O. Sjaastad, A. Langslet, I. Sulg, K. Egge, J. Diderichsen, A new syndrome: thrombocytopathia, muscle fatigue, asplenia, miosis, migraine, dyslexia and ichthyosis, Clin. Genet. 28 (5) (Nov 1985) 367–374.
[6] J. Böhm, F. Chevessier, A. Maues De Paula, C. Koch, S. Attarian, C. Feger, D. Hantaï, P. Laforêt, K. Ghorab, J.M. Vallat, M. Fardeau, D. Figarella-Branger, J. Pouget, N.B. Romero, M. Koch, C. Ebel, N. Levy, M. Krahn, B. Eymard, M. Bartoli, J. Laporte, Constitutive activation of the calcium sensor STIM1 causes tubular-aggregate myopathy, Am. J. Hum. Genet. 92 (2) (Feb 7 2013) 271–278.
[7] C. Hedberg, M. Niceta, F. Fattori, B. Lindvall, A. Ciolfi, A. D'Amico, G. Tasca, S. Petrini, M. Tulinius, M. Tartaglia, A. Oldfors, E. Bertini, Childhood onset tubular aggregate myopathy associated with de novo STIM1 mutations, J. Neurol. 261 (5) (May 2014) 870–876.

Amrathlal Rabbind Singh*
Gilles Morin
Jacques Rochette
EA 4666 and Department of Molecular and Clinical Genetics, CHU d'Amiens, Université de Picardie Jules Verne, 80054 Amiens, France
*Corresponding author.
E-mail address: rabbind@gmail.com (A.R. Singh).

22 January 2015

http://dx.doi.org/10.1016/j.ymgmr.2015.01.003