Mahmut Çerkez Ergören, Sehime Gülsün Temel*[1]
Departments of Medical Biology, *Embryology and Histology, Faculty of Medicine, Near East University, Near East Boulevard; Nicosia-*Turkish Republic of Northern Cyprus*
[1]Department of Embryology and Histology, Faculty of Medicine, Uludağ University; Bursa-*Turkey*

References

1. Baydar CL, Özen M. A case of hypertrophic and dilated cardiomyopathic sudden cardiac death: *de novo* mutation in *TTN* and *SGGD* genes. Anatol J Cardiol 2016 Jul 31. Epub ahead of print. Crossref
2. Veltman JA, Brunner HG. *De novo* mutations in human genetic disease. Nat Rev Genet 2012; 13: 565-75. Crossref
3. http://exac.broadinstitute.org
4. Pugh TJ, Kelly MA, Gowrisankar S, Hynes E, Seidman MA, Baxter SM, et al. The landscape of genetic variation in dilated cardiomyopathy as surveyed by clinical DNA sequencing. Genet Med 2014; 6: 601-8. Crossref
5. General Variant Classification Assertion Criteria. GeneDx DNA Diagnostic Experts. http://www.genedx.com

Address for Correspondence: Dr. Mahmut Çerkez Ergören
Department of Medical Biology, Faculty of Medicine
Near East University, Near East Boulevard
Nicosia-*Turkish Republic of Northern Cyprus*
E-mail: mahmutcerkez@gmail.com
©Copyright 2017 by Turkish Society of Cardiology - Available online at www.anatoljcardiol.com
DOI:10.14744/AnatolJCardiol.2017.7554

Author`s Reply

To the Editor,

We are delighted with the interest shown in our work.

In our article published in the Anatolia Journal of Cardiology in late 2016 entitled "A hypertrophic and dilated cardiomyopathic sudden cardiac death case; de novo mutations TTN and SGCD genes", we demonstrated that the likely benign [NM_000337.5(SGCD):c.15G>C:(p.Glu5Asp)] and the missense [NM_003319.4(TTN):c.21758T>C (Ile7253Thr)] variants could be associated with dilated cardiomyopathy (DCM)/hypertrophic cardiomyopathy (HCM) as a (cor bovinum) disease or in young sudden cardiac death (1).

In the letter to the editor, it was claimed that our variant (TTN):c.21758T>C had been previously identified by Pugh et al. (2) and that therefore, the variant is no longer novel. However, this is immaterial because there is no such variant (TTN):c.21758T>C reported by Pugh et al. (2) (see Suplementary-1 Cases).

Although the genes NM_000337.5(SGCD):c.15G>C (p.Glu5Asp) and NM_003319.4(TTN):c.21758T>C (Ile7253Thr) were identified as likely benign and missense variants, respectively, in National Center for Biotechnology Information (NCBI) database and had not been previously reported as disease or death-causing variants, we found that titin (TTN) and sarcoglycan (SGCD) genes are associated with HCM/DCM and DCM, since cause of death was determined to be sudden circulatory failure resulting from DCM/HCM.

We used the term "de novo" in our case report to mean a new instance, and perhaps were not attentive enough to its very specific genetic nomenclature. Regarding the comments on SGCD variant of "unknown significance," there are many instances of single point mutations causing serious disease (e.g., sickle cell anemia). While we cannot definitively conclude that the mutation caused the heart pathology, we believe it is important to report this and similar cases, as these are relevant to whether these variants could merit further study. We agree that larger cardiologic clinical studies and sophisticated genetic studies carried out by specialists are required to clarify these issues. However, this lies outside the scope of the current work.

Çetin Lütfi Baydar
Department of Forensic Medicine, Faculty of Medicine, Near East University; Nicosia-*Turkish Republic of Northern Cyprus*

Reference

1. Baydar ÇL, Özen M. A hypertrophic and dilated cardiomyopathic sudden cardiac death case; de novo mutations TTN and SGCD genes. Anatol J Cardiol 2016 Jul 31. Epub ahead of print. Crossref
2. Pugh TJ, Kelly MA, Gowrisankar S, Hynes E, Seidman MA, Baxter SM, et al. The landscape of genetic variation in dilated cardiomyopathy as surveyed by clinical DNA sequencing. Genet Med 2014; 6: 601-8. Crossref

Address for Correspondence: Dr. Çetin Lütfi Baydar
Near East University, Faculty of Medicine
Department of Forensic Medicine, Nicosia-*Turkish Republic of Northern Cyprus*
E-mail: drcetinlutfi@yahoo.com

Role of ABO blood groups in prosthetic valve thrombosis

To the Editor,

We read with great interest the article published in Anatolian Journal of Cardiology by Astarcıoğlu et al. (1) entitled "ABO blood types: impact on development of prosthetic mechanical valve thrombosis." Several risk factors of prosthetic valve thrombosis (PVT) are well known. The search for new categories of risks should continue to refine even more the initial therapeutic decision in PVT. In this work, the authors evaluated the association between blood group status and PVT. They reported that patients with non-O blood groups have greater incidence of PVT compared with O blood groups. This result suggests that non-O group may be a risk factor that favors developing PVT.

It is increasingly recognized that individuals with non-O blood groups may be at elevated risk of venous and arterial thromboembolic events compared with individuals with blood group O. This increased risk has been attributed to higher concentrations of factor VIII and von Willebrand factor (2).