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Short Communication

Precision Medicine of Channelopathies: The successful model of Long QT syndrome

Ghayath Baroudi

Institute for genetics and channelopathies, Quebec, Canada

Correspondence

Ghayath Baroudi, PhD

Institute for genetics and channelopathies; 111, Route 350, Sainte-Ursule, Quebec, J0K 3M0, Canada

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A few years ago, the notion of "precision medicine" was defined by the American National Research Council in their Precision Medicine Initiative as an emerging approach for disease treatment that takes into account individual variability in genes, environment and lifestyle. Since then, it has rapidely expanded over the former notion of "personalized medicine" which may ambiguously indicate the design of drugs or devices to treat litterarly every unique patient differently. Precision medicine rather indicates subcategorizing patients into homogeneous groups in order to offer them a better management of disease and provide more efficient therapeutic approaches.

Indeed, the genetic profile of individuals is a major determinant in the process of patients subcategorization. Generally, the pathophysiology of diseases involves distinct cellular and physiological mechanisms in individuals who have different genetic profils. Therefore, it was essential for precision medicine practicioners to consider genetic nuances of each patient prior to tailoring any therapeutic intervention.

Channelopathies represent a well-defined class of diseases that are caused essentially by abnormal ion channels. Mainly, ion channels abnormalities result from subtile alterations at the genetic level. Mutations in genes encoding ion channels or their regulatory subunits lead to the formation of dysfunctional channels that are at the very basis of the pathophysiology of channelopathies. Interestingly, channelopathies regroup a wide variety of diseases including cardiac, skeletal muscle and nervous system diseases. Each channelopathy is caused by one or more mutations that may occur individually or combined, in one or more genes.

Throughout the last decade, a huge number of mutations causing channelopathies have been identified and characterized. Interestingly, extensive research from multiple laboratories characterizing ion channel mutants have unravelled contrasting impacts of mutations on the pathophysiology of channelopathies. Whereas some mutations were shown to cause abnormal gain of function to ion channels, others reduces channels function or even abolish it completely. Accordingly, this required differential therapeutic approaches to be administered. A representative example of precision medicine application in disease management is that of cardiac channelopathies.

Long QT syndrome, a cardiac arrhythmogenic channelopathy associated with malignant arrhythmia and risk of sudden death, results from the occurrance of a wide variety of mutations mainly in genes encoding sodium, potassium and calcium channels or their regulatory subunits. The functional impact that each mutation exerts on ion channels is diverse and sometimes opposite to that caused by other mutations leading to the very same condition. Consequenlty, long QT syndrome has been subclassified into 17 distinct categories depending on the dysfunctional gene, and hence, on the patient genetic profil. Accordingly, therapeutic aproaches are then applied to patients in a precise and more personalized manner. Interestingly, the application of precision medicine the way it is administered with LQTS patients has proven to reduce the mortality rate from 71% in 1985 [1] to only 0.3% in our days [2].

Indeed, current advances in molecular technologies and their systematic implementation as essential components in routine practices have revolutionized the way many diseases are diagnozed and managed. Combined with the emerging targeted therapeutic techniques, this can take the mission of precision medicine very far in attaining its objective of providing complete protection to every individual.

References

1. Rohatgi RK, Sugrue A, Bos JM, et al. Contemporary outcomes in patients with long QT syndrome. J Am Coll Cardiol. 2017;70(4):453–462.
1. Schwartz PJ. Idiopathic long QT syndrome: progress and questions. Am Heart J. 1985;109(2):399–411.

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