Abstract

Health Science Journal
ISSN 1791-809X

2023
Vol.17 No.P53

Genetics: cross and delight for a modern internist

Savino I.G[1], Cammarota A[2], Lepre D[2], Ronga I[1], Barbieri V[1], Bova M[1], Iannuzzi A[1]

[1]Department of Internal Medicine AORN Cardarelli

[2]Department of Internal Medicine A.O.U. Federico II

Background

The great advances in the development of genomic technologies and their incorporation into routine clinical practice is bringing new astonishing challenges for internists. At present, the traditional terms ''mutation'' or "polymorphism" are being replaced by the term ''variant''. The interpretation of genetic test is still a challenge, in some cases leading to misdiagnosis and unwarranted diagnostic investigations.

Case History

We describe a case of suspect acute hepatic porphyria previously diagnosed as fibromyalgia. A 55 years old woman since the age of thirty, suffered from generalized pain, limb paraesthesia's and recurrent severe and diffuse abdominal pain, poorly localized, requiring access to emergency department. The patient had also vesicular skin lesions on the body's areas exposed to sunlight, previously diagnosed as urticarial.

Biochemical test showed mild hyponatremia and abdominal ultrasound and CT scan were unremarkable. We dosed ALA and PBG on a light-shielded urine sample and a genetic test was performed for suspect porphyria .The results described variants of unknown significance (VUS, class III) in a gene involved in porphyria, making it difficult to establish the genotype-phenotype association. Bio molecular investigations are still in progress.

Discussion

Based on our experience, and on the clear necessity of incorporation of genomic technologies into routine clinical practice, we propose to implement and strengthen the education and training of internists on genomic medicine, and to create multidisciplinary teams to manage difficult cases.

Bibliography

1. K J Hofman, E S Tambor, G A Chase, G Geller, R Faden N A (1993) Holtzman Physicians' knowledge of genetics and genetic tests, Acad Med 68:625-32.
2. Bruce Wang, Herbert L, Bonkovsky, Joseph K Lim, Manisha Balwani (2023) AGA Clinical Practice Update on Diagnosis and Management of Acute Hepatic Porphyrias: Expert Review. Gastroenterology. Mar 164: 484-491.