Review Article **AJODRR (2018) 1:1**

American Journal of Dermatological Research and Reviews
(DOI:10.28933/AJODRR)

Phenotype / genotype relationship of Darier's disease

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ABSTRACT

Introduction - Darier's disease is a severe hereditary keratinization disorder that is transmitted by an autosomal dominant factor of an unusual type of genodermatosis. The age of onset of symptoms is typically at puberty and equally affects both sexes. Objectives - Cytogenetically show the behavior of the disease in individuals, its clinical and histopathological characteristics. Contribute to a better understanding of the phenotype / genotype relationship of the disease. Methodology - This literature review on the disease was carried out with information searches in Dermatology books, Oral Pathology and published scientific articles. Results - It is believed that the disease is related to a gene, ATP2A2, which is responsible for coding a calcium pump. From this, studies show that some cellular structures, such as desmosomes, are compromised (because chemical signaling depends on the intracellular calcium), with a lack of cohesion between epithelial cells. Conclusion - The phenotype / genotype relationship of this disease is not yet fully understood and the disease represents a challenge for medical genetics.

Keywords: Pump ; Calcium ; Disease

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How to cite this article:
Monteiro, P. S. R.; Macedo, A. E., Mallemont, L. B., Silva, I. C., Silva, S. R. G. Santana Arruda, H. S.Phenotype / genotype relationship of Darier's disease.American Journal of Dermatological Research and Reviews, 2018, 1:1

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