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CARDIO-FACIO-CUTANEOUS SYNDROME, A RARE DISEASE: LITERATURE REVIEW

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INTRODUCTION

Cardio-Facio-Cutaneous Syndrome (CFC) is a rare genetic disease characterized by multiple congenital anomalies and intellectual disability, congenital heart defects, the most common being pulmonary stenosis and atrial septal defect, as well as ectodermal abnormalities and growth deficiency. They have developmental delay and intellectual disability, usually ranging from moderate to severe. Typical facial features are a high forehead with bitemporal constriction, hypoplasia of the supraorbital ridges, antimongoloid inclination of the palpebral fissures, a depressed nasal bridge and posteriorly angled ears with prominent helices. The hair is generally sparse and friable. Skin changes ranged from irregular hyperkeratosis to a condition similar to severe generalized ichthyosis. Differential diagnosis should be made with Noonan and Costello Syndrome as the signs and symptoms are similar and can be distinguished by their genetic cause and by some specific patterns of signs and symptoms.

OBJECTIVE

To analyze the general characteristics of CFC Syndrome, including clinical manifestations, diagnosis and treatment.

METHODOLOGY

The research was carried out in the Lilacs, VHL and Pubmed databases between 2014 and 2022. The descriptors used were "syndrome", "cardio-facio-cutaneous" and their English counterparts, "syndrome" and "cardio--facio-cutaneous". Twenty articles were found, 13 of which were excluded because they did not specifically address the disease studied.

RESULTS AND DISCUSSION

Of the seven articles analyzed, five showed that CFC syndrome is inherited in an autosomal dominant manner, with the known mutations found in the KRAS, BRAF, MAP2K1 and MAP2K2 genes being responsible for the characteristics of the disease. There are also some individuals who do not have a mutation in any of these genes but do have the syndrome. The expression of the phenotypes that result in clinical manifestations depends on how the genes are expressed.

CONCLUSION

CFC Syndrome does not have a definitive cure and its treatment is only symptomatic with special and occupational education, speech therapy and appropriate skin care. Surgical intervention is often successfully used for symptomatic relief in people with obstructive cardiomyopathy, and in rare cases heart transplantation may be chosen. Periodic echocardiograms are essential to check for possible hypertrophic cardiomyopathy. Therefore, it is necessary to use genetic analysis techniques to detect mutations, especially in the BRAF gene. Each specific sign and symptom should be treated appropriately for a better quality of life.

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