Pharmacogenomics and Personalized Medicine

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LETTER

A Case Report of Cardiofaciocutaneous Syndrome with MAP2K1 Pathogenic Variant [Letter]

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Dear editor

With all due respect, we have read the article published by Qiong Tang et al in the Pharmacogenomics and Personalized Medicine and it would be good if there were suggestions that could inspire further research. The author describes a case report of a child aged two years and six months who experienced Cardiofaciocutaneous Syndrome.[1] Cardiofaciodermal Syndrome is included in RASopathies, namely conditions caused by treatment of the gene that codes for the mouse sarcoma pathway protein/mitogen-activated protein kinase (RAS/MAPK). Clinical manifestations are facial dysmorphism, growth failure, cardiac disorders, developmental delay, and ectodermal abnormalities. Apart from that, the cause is mutations in four genes, namely BRAF, MAP2K1, MAP2K2, and KRAS).[2,3]

The genetic examination carried out on these patients was an examination of the MAP2K1 gene, which according to previous research, this gene has a small percentage as the cause of Cardiofaciodermal Syndrome with an average of 25%. The input we provide is to check all participating genes, namely BRAF, MAP2K2, and KRAS. By examining these three genes, it is hoped that we can find out which gene plays a more important role in this case. The gene variants that are often the cause are the BRAF gene (75%), MAP2K1 and MAP2K2 (25%) and KRAS (2%) which is a new pathogen whose incidence rate is still rarely found.[4,5]

Examination of clinical symptoms and genetic examination in cases of Cardiofacioquantum Syndrome must be carried out completely in order to obtain accurate results. If in certain cases there are unclear clinical symptoms, it is recommended that genetic examination be carried out to see whether there are mutations in the genes that play a role.[6] Research into this case must continue to be carried out if there are new cases related to this disease.

Acknowledgments

Thank you to all authors who have provided assistance and support during the review process and writing the article. We would like to express our gratitude to Dr. Sunarno for his continuous support and valuable input during the writing of this manuscript.

Disclosure

The authors report no conflicts of interest in this communication.

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Received: 30 September 2023
Accepted: 16 October 2023
Published: 25 October 2023

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https://doi.org/10.2147/PGPM.S442628