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Clinical and molecular genetic findings of a patient with fucosidosis: A case report

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Fucosidosis is an autosomal recessive lysosomal storage disease caused by defective alpha-L-fucosidase with accumulation of fucose in the tissues. Clinical features include angiokeratoma, progressive psychomotor retardation, neurologic signs, coarse facial features, hepatomegaly, and dysostosis multiplex. Fucosidosis is caused by disruptions or changes (mutations) of the alpha-L-fucosidase (FUCA1) gene resulting in deficiency of the alpha-L-fucosidase enzyme. The symptoms of fucosidosis occur as a result of excessive accumulation of fucose-containing compounds (e.g., certain glycosphingolipids or certain glycoproteins) in the body due to abnormally low levels of this enzyme. The aim of this study is to present clinical and molecular genetic findings of a patient with fucosidosis. Four year old Caucasian boy brought by parents to the Istanbul University, Faculty of Medicine, Department of Urology because of widespread angiokeratoms on his skrotums. The clinical examination revealed short stature, coarse facial features, bilateral ptosis, hypertonic lower extremities, psychomotor retardation and asthma. The asthma symptoms has been managed in the Department of Pulmonary Medicine since 2,5 years. The oral examination revealed mouth breathing and hypertrophic gums. The MR findings and angiokeratoms indicated fucosidosis, the patient directed to the Bezmialem Vakif University Department of Medical Genetics and the DNA sequence analysis about *FUCA1* showed Q82Ter missense mutation. The patient was the child of a consanguineous parent parent. The genetic tests must be implemented in such families before pregnancy.

Biography

Faruk Ozcan completed her PhD in Department of Urology at Istanbul University in 1991. He has published more than 30 papers in reputed journals and has advised undergraduate and PhD thesis. He has been working as a Professor in Department of Urology at Istanbul University since 2000.

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