

A patient with ascending aortic dilatation, similar to phenotypes of connective tissue disorders

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Genet. Mol. Res. 8 (2): 426-434 (2009)

Received February 15, 2009

Accepted March 6, 2009

Published April 14, 2009

ABSTRACT. We report on the clinical and molecular findings of a patient who presented alopecia, epicanthus, micrognathia, retrognathia, high arched palate, hypertelorism, Chiari type I malformation, mixed-type hearing loss but with normal heartbeat Q-T interval, malformed earlobes, down-slanted palpebral fissures, downturned corners of the mouth, syndactyly, atopic eczema, and seizures. The patient was a male adult, 23 years old, with short stature (153 cm) and low weight (50.5 kg), due to severe aortic insufficiency and dilatation of the ascending aorta. Conventional cytogenetic screening did not show any chromosomal gains or losses. Molecular genetic screening was conducted for gene mutations involved in various syndromes; the mutations found included [β -fibrinogen -455 G>A wt/wt (wt/mut), PAI-1 4G/5G (4G/4G), HPA1 a/b (a/a), MTHFR C677T wt/wt (wt/mut),

ACE I/D (I/I), and Apo E E3/E4]. Many clinical and molecular genetics findings overlapped with other conditions associated with arterial tortuosity and arterial aneurysms, including the Marfan, Ehler-Danlos, Shprintzen-Goldberg, and Loeys-Dietz syndromes. Although a diagnosis of Shprintzen-Goldberg syndrome was based on clinical findings and radiographic findings indicate other syndromes, aortic root dilatation seems to be a new symptom, similar to phenotypes of connective tissue disorders. The unique grouping of clinical manifestations in this patient and the molecular genetics findings lead us to suggest that this case could be an example of a previously unrecognized syndrome.

Key words: Mimicking; Ascending aorta; Connective tissue disorders; Phenotypes