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Letter to the editor

R298Q mutation of *p63* gene in autosomal dominant ectodermal dysplasia associated with arrhythmogenic right ventricular cardiomyopathy

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Abstract

Mutations in the *p63* gene have been identified in five types of syndromic ectodermal dysplasias (EDs) with overlapping phenotypes: Ectrodactyly–Ectodermal dysplasia–Clefting (EEC syndrome, MIM 604292), Ankyloblepharon–Ectodermal dysplasia–Clefting (AEC syndrome, MIM 106260) [3], Acro-Dermato-Ungueal-Lacrimal-Tooth (ADULT syndrome, MIM 103285), Rapp–Hodgkin (RHS syndrome, MIM 129400) and Limb–Mammary (LMS syndrome, MIM 603543) [2].

In all those conditions congenital heart defects have been only occasionally found and to date, arrhythmogenic right ventricular dysplasia/cardiomyopathy (ARVC) has never been observed in patients affected by *p63*-related ectodermal dysplasia [9]. Here we describe for the first time this association.

Keywords: *p63*; Arrhythmogenic right ventricular cardiomyopathy; Ectodermal dysplasia

1. Case report

A 17-year-old boy was referred to medical attention because of asthenia, dyspnea and cardiopalm. At clinical examination he had normal growth parameters. Ectodermal signs such as hypodontia with enamel and dentine dysplasia, persistence of four deciduous teeth, dystrophic nails, sparse, fragile and wiry hair, decreased sweating and right atelia were seen (Figs. 1 and 2).

Microscopic hair examination showed trichorrhexis nodosa. He did not have significant skin changes such as dry skin, freckling or photosensitivity. Hands and feet were clinically normal and nor radiographic anomalies were seen. Ocular abnormalities included lacrimal duct aplasia and recurrent blepharitis. In the first months of life this boy had suffered from many episodes of hyperpyrexia during summer probably due to reduced sweating.

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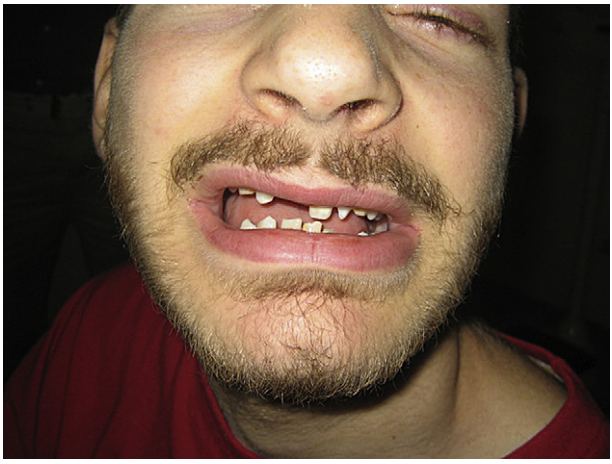

Fig. 1. Hypodontia with enamel and dentine dysplasia, persistence of deciduous teeth.

Cardiological findings were consistent with the diagnosis of ARVC by morphological, functional, ECG and histologic features. Following amiodarone therapy he developed hypothyroidism and L-tiroxine treatment was started. A cardioverter defibrillator was implanted one year after diagnosis.

His mother had atelia, hypodontia and currently she wears dentures but she had no heart defects or arrhythmias. Mother's echocardiography and EKG were negative; the maternal family history was negative for sudden death, rhythm disturbances and dilated cardiomyopathy.

2. Discussion

ARVC is most often an autosomal dominantly inherited cardiomyopathy with primarily right ventricular involvement. The clinical presentation is highly variable but common findings are ventricular arrhythmias, syncope, and sudden death. Diagnosis is based on internationally

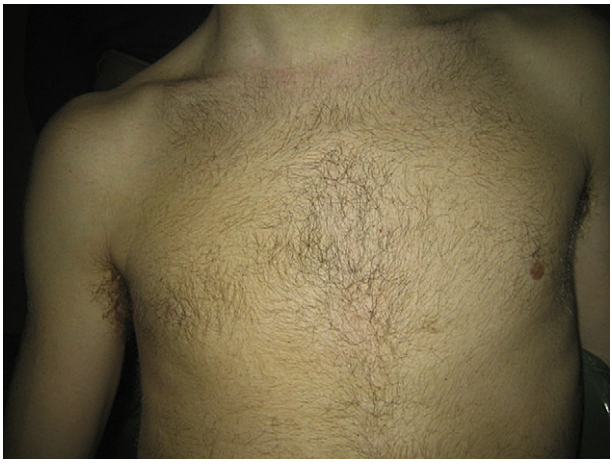

Fig. 2. Atelia.