

POSTER PRESENTATION

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Phenotypic variability in a cohort of 40 Italian subjects carrying mutations in the gene *EDA*

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Ectodermal dysplasias (ED) are a group of clinically and genetically heterogeneous conditions commonly characterized by abnormal development of at least two structures derived from the embryonal ectoderm (hair, teeth, nails, and sweat glands). X-linked hypohidrotic ED, which is caused by mutations in the gene EDA (MIM 305100), is the most frequent form. In this study, we investigated the phenotype of 40 male patients, aged 2 to 20 years, who all showed developmental defects of ectodermal derivatives and a mutation in EDA. Specialist assessments of the involved organ systems were performed. 95% of these patients presented with impairment of sweating, which was only moderate in 35%, while two subjects did not show any alteration of sweat gland function. Severe oligodontia was found in 80% of the subjects, 10% had hypodontia. 90% of the patients showed abnormal crown morphology of the teeth. Severe involvement of the scalp hair was observed in 22% of the patients, moderate involvement in 72%, and no relevant alterations of hair morphology, quantity or growth in 2 patients. Onychodystrophy was seen in 67% of our patients. Concerning minor alterations of ectodermal tissues, we found dry eye signs in 92% of the subjects investigated, recurrent respiratory infections in 82%, hearing loss in 10%, atopic dermatitis in 35%, and a neuropsychological disorder in 10%. Our study shows that an EDA mutation can be present in males also in the context of only one of the major clinical signs of Xlinked ED, but associated with minor alterations. Therefore we suggest that EDA gene analysis should be considered also for males with a mild ED phenotype.

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