TP63-related disorders (ectrodactyly-ectodermal dysplasia-clefting [EEC3] syndrome; acro-dermato-ungual-lacrimal-tooth [ADULT] syndrome; ankyloblepharon-ectodermal dysplasia-clefting [AEC; hay-wells] syndrome; limb-mammary syndrome (LMS); nonsyndromic split-hand/split-foot malformation [SHFM4]; rapp-hodgkin syndrome [RHS])

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Citation
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Authors' conclusions
Ectodermal dysplasias (EDs) occur in approximately 7 of 10,000 births and are characterized by abnormal development of hair, teeth, nails, sweat glands, or skin. Ectrodactyly-ectodermal dysplasia-clefting syndrome (EEC3) is the prototype of a distinct group of EDs that have been associated with variants in the tumor protein p63 (TP63) gene. Clinical characteristics of EEC3 are variable in severity but include three major categories: evidence of ED such as sparse or absent hair and skin dysplasia; limb malformations such as absence or fusion of digits in the hand or foot; and orofacial underdevelopment and/or clefting. The other disorders associated with TP63 variants are defined by the presence of characteristics from at least one of the three phenotypic categories seen in EEC3. These disorders are acro-dermato-ungual-lacrimal-tooth (ADULT) syndrome; ankyloblepharon-ectodermal dysplasia-clefting (AEC; Hay-Wells) syndrome; limb-mammary syndrome (LMS); and nonsyndromic split-hand/split-foot malformation (SHFM4). Cases of Rapp-Hodgkin syndrome (RHS), also known to be associated with TP63 variants, are now included within the spectrum of AEC. Tumor protein p63 is a transcription factor important to cell regulation, instrumental in development of ectodermal structures such as teeth, skin, nails and hair, the urinary system, limbs, and facial structure. TP63-related disorders occur in females and males, and may be inherited in an autosomal dominant manner or may occur spontaneously in patients with no family history of disease. Specific characteristics may vary between related individuals, indicating clinical variability associated with TP63 gene variants, or the potential for other unknown genetic or environmental factors of influence. The molecular pathogenesis of TP63-related disorders is not well understood. Treatment for TP63-related disorders is symptom driven and consists of wound care and infection prevention, wigs, and dentures and/or dental implants. Appropriate therapies and surgical treatment of orofacial clefting and limb malformations may also be incorporated into treatment.

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