Congenital generalized hypertrichosis terminalis: a proposed classification and a plea to avoid the ambiguous term "Ambras syndrome".

Abstract:
Congenital generalized hypertrichosis terminalis (CGHT) is a heterogeneous group of diseases with continuing excessive growth of terminal hair. "Ambras syndrome" was first coined by Baumeister in 1993 to describe a case of nonsyndromic CGHT which was erroneously analogized to the portrait paintings of Petrus Gonzales and his children, exhibited in Ambras Castle near Innsbruck, Austria. This family probably, a syndromic type with abnormal dentition, inherited as an autosomal dominant trait. CGHT associated with gingival hyperplasia is probably a particular entity typified by the historical cases of Julia Pastrana and her son. An X-linked type of CGHT has likewise been categorized as "Ambras syndrome". Moreover, some reports have mistakenly classified "Ambras syndrome" as an example of hypertrichosis lanuginosa. Potential gene loci identified so far may include 8q22, 17q24.2-q24.3 and Xq24-q27.1. The designation "Ambras syndrome" has thus been applied to various types of congenital hypertrichosis that differ to such degree that the name "Ambras" has no specific meaning, neither in the past nor in the future. Hence, this misleading term should now be jettisoned.

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