Opinion

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Rapp-Hodgkin Ectodermal Dysplasia Syndrome

Yakubu OE*

Federal University Wukari, PMB 1020, Katsina Ala Rd, Nigeria

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***Corresponding author:**
Dr. Ojochenemi Ejeh Yakubu

oj4real_2007@yahoo.co.uk

Federal University Wukari, PMB 1020, Katsina Ala Rd, Nigeria.

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Opinion

Rapp-Hodgkin syndrome is a rare disease that can affect your hair, nails, skin, sweat glands, and teeth. It is caused by a problem with your genes, and it is part of a larger group of conditions that doctors call ectodermal dysplasia. Rapp-Hodgkin syndrome has signs and symptoms that overlap extensively with those of ankyloblepharon-ectodermal defects-cleft lip/palate (AEC) syndrome, also called Hay-Wells syndrome.

These two syndromes were classified as separate disorders until it was discovered that they both result from mutations in the same part of the same gene, *TP63*. Most researchers now consider Rapp-Hodgkin syndrome and AEC syndrome separate disorders that are part of the same disease spectrum. Several forms of ectodermal dysplasias with overlapping clinical features have been found to be due to mutations in p63, a transcription factor that is structurally related to p53, localized to gene locus 3q27. These include Rapp–Hodgkin syndrome, ectodermal dysplasia, and cleft lip/palate syndrome 3; ankyloblepharon–ectodermal defects–cleft lip/palate syndrome, limb–mammary syndrome, and nonsyndromic split-hand/foot malformation are caused by mutations in p63 as well.

Rapp-Hodgkin syndrome (RHS) is characterized by the anhidrotic ectodermal dysplasia and cleft lip/palate. Patients have characteristic facies, wiry, slow growing, and uncombable hair, obstructed lacrimal puncta/epiphora, bilateral stenosis of external auditory canals, microsomia, hypodontia, cone-shaped incisors, enamel hypoplasia, and dystrophic nails. A condition known as Rapp-Hodgkin syndrome has signs and symptoms that overlap considerably with those of AEC syndrome. These two syndromes were classified as separate disorders until it was discovered that they both result from mutations in the same part of the same gene.

Ankyloblepharon-ectodermal defects-cleft lip/palate (AEC) syndrome is caused by mutations in the *TP63* gene. This gene provides instructions for making a protein known as p63, which plays an essential role in early development. The p63 protein is a transcription factor, which means that it attaches to DNA and controls the activity of particular genes. The p63 protein turns many different genes on and off during development. It appears to be especially critical for the development of ectodermal structures, such as the hair, skin, teeth, and nails. Studies suggest that it also plays important roles in the development of the limbs, facial features, urinary system, and other organs and tissues. The *TP63* gene mutations responsible for AEC syndrome interfere with the ability of p63 to turn target genes on and off at the right times.

It is unclear how these changes lead to abnormal ectodermal development and the specific features of AEC syndrome. When genetic testing became available, this suspicion was confirmed by the finding that HWS and RHS were both caused by mutations in the same domain of the p63 gene. Identical p63 gene mutations have been detected in patients diagnosed with either HWS or RHS. Consequently, these ectodermal dysplasias are now considered the same disorder, which is now referred to as ankyloblepharon-ectodermal defects-cleft lip and palate AEC syndrome.

