

A CASE REPORT

HAY-WELLS SYNDROME OF ECTODERMAL DYSPLASIA

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ABSTRACT

Hay-Wells syndrome is a rare form of ectodermal dysplasia initially described by Hay and Wells in 1976. It is an autosomal dominant disorder with varying forms of expression featuring congenital abnormalities of the skin, hair, teeth, nails and sweat glands. Reporting a case with the clinical features of cleft lip, cleft palate, ankyloblepharon, multiple synchia in oral cavity, dysplasia of toe nails and dry peeling skin.

Keywords: Hay-Wells syndrome, Ectodermal dysplasia

1.INTRODUCTION

Ankyloblepharon-ectodermal dysplasia-cleft lip/palate (AEC) syndrome, which is also known as Hay-Wells syndrome, is a rare disorder characterized by a wide variety of symptoms that can affect the skin, hair, nails, teeth, certain glands, arms and legs.

This is rare genetic disorder in humans, initially described by Hay and Wells in 1976 in seven individuals from 4 families in whom multiple and complex malformations were associated with a pattern of autosomal dominant inheritance of varying degrees of penetrance ⁽¹⁾.

Most authors consider the presence of ankyloblepharon filiforme adnatum (tissue strands joining the upper and lower eyelid margins), cleft lip and/or palate and findings consistent with ectodermal dysplasia to constitute essential criteria for a diagnosis of Hay-Wells syndrome. Common ectodermal defects include alopecia, onychodystrophy, oligodontia and hypohydrosis

AEC syndrome is caused by mutations in the p63 gene ⁽²⁾, and most cases are either new (spontaneous) mutations or are inherited as autosomal dominant disorders. Another disorder that is caused by mutations in the p63 gene, Rapp Hodgkin syndrome, is now considered to be part of the one disease spectrum that also includes AEC syndrome.

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2.CASE REPORT

Presenting a case report of a new born male baby born to a consanguineous parents. On examination the baby had clinical features of cleft lip, cleft palate, ankyloblepharon, multiple synchia in oral cavity, dysplasia of toe nails and dry peeling skin.

Laboratory tests including complete blood count, electrolytes and liver, kidney and thyroid function tests showed no abnormalities. Chest radiography, cardiac ECHO and USG abdomen done to rule out other anomalies was essentially normal

Multiple synchia in oral cavity and ankyloblepharon was released by cauterization.

3.DISSCUSSION

Hay-Wells or AEC syndrome is a rare, autosomal dominant disease. Ankyloblepharon filiforme adnatum, ectodermal dysplasia and cleft lip and/or palate are considered cardinal signs of this syndrome by most authors ⁽³⁾. All these features were present in the case reported here.

Ankyloblepharon consists of partial or complete fusion of the upper and lower eyelid margins, and is often sporadic. Normally, the eyelids remain fused until the fifth week of pregnancy at which time they separate. Therefore, anomalies that occur between the 7th and 15th weeks of pregnancy may result in palpebral abnormalities.⁽⁴⁾ Ankyloblepharon may also be present in trisomy 18 and CHAND (Curly Hair-

Ankyloblepharon-Nail Dystrophy) syndrome, and is associated with cardiac defects, hydrocephaly, imperforate anus and glaucoma; therefore, its presence should always serve as an alert to the possibility of another important concomitant disorder.



Ectodermal dysplasias refers to a group of diseases in which defects occur in the development of the hair, teeth, nails, sweat glands and other structures originating from the ectoderm.^(5,6) These abnormalities, when associated with other malformations, constitute a group of ectodermal dysplasia syndromes that includes the ectrodactyly-ectodermal dysplasia-clefting (EEC) syndrome, Rapp-Hodgkin syndrome and the CHAND syndrome, which represent the principal differential diagnoses for the presently reported syndrome.⁽⁶⁾ Patients with Hay-Wells syndrome may present various degrees of alopecia, sparse, fair hair, onychodystrophies, palmoplantar hyperkeratosis, skin pigmentation disorder,⁽⁷⁾ hypohidrosis, hypodontia, dental malformations and auricular deformities,⁽⁸⁾ all of which were present in the patient in question. Lacrimal duct obstruction is a common finding. Other reported findings include supernumerary nipples, otitis media, hypospadias, midfacial hypoplasia, hypertelorism, low stature, intellectual impairment, hypoacusia and ocular abnormalities.

The importance of the early diagnosis of this syndrome should be emphasized in order to implement appropriate genetic counselling for parents, as well as clinical and dermatological treatment of the patient at the initial phase of erythroderma and management of scalp infections. In addition, ophthalmological and odontological care should be initiated and cleft lips and palates corrected surgically.

4.REFERENCES

1. Hay RJ, Wells RS. The syndrome of ankyloblepharon, ectodermal defects, and cleft lip and palate: an autosomal dominant condition. *Br J Dermatol.* 1976;94:277-89.
2. Payne AS, Yan AC, Ilyas E, Li W, Seykora JT, Young TL, et al. Two novel TP63 mutations associated with the ankyloblepharon, ectodermal defects, and cleft lip and palate syndrome - a skin fragility phenotype. *Arch Dermatol.* 2005;141:1567-73.
3. Macias E, de Carlos F, Cobo J. Hay-Wells syndrome (AEC): a case report. *Oral Dis.* 2006;12:506-8.
4. Mohamed YH, Gong H, Amenyra T. Role of apoptosis in eyelid development. *Exp Eye Res.* 2003;76:115-23.
5. Tsutsui K, Asai Y, Fujimoto A, Yamamoto M, Kubo M, Hatta N. A novel p63 sterile alpha motif (SAM) domain mutation in a Japanese patient with ankyloblepharon, ectodermal defects and cleft lip and palate (AEC) syndrome without ankyloblepharon. *Br J Dermatol.* 2003;149:395-9.
6. Pinheiro M, Freire-Maia N. Ectodermal dysplasias: a clinical classification and a causal review. *Am J Med Genet.* 1994;53:153-62.
7. Drut R, Pollono D, Drut RM. Bilateral nephroblastoma in familiar Hay-Wells syndrome associated with familial reticulate pigmentation of the skin. *Am J Med Genet.* 2002;110:164-9.
8. Kulkarni ML, Deshmukh S, Matani D, Gayatri K. Hay- Wells syndrome of ectodermal dysplasia. *Indian J Pediatr.* 2006;73:101.
