

ARCHIVES OF DISEASE IN CHILDHOOD

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Annotation

Ectodermal dysplasia

Ectodermal dysplasia is a very rare condition occurring in an estimated one per 100 000 live births.¹ It can be associated with early morbidity and mortality.² The disease was first described by Thurnman in 1843.³ It embraces a long series of abnormalities whose common denominator is a morphological alteration of ectoderm derived organs and tissues.⁴ Structures most commonly involved are the eccrine glands (resulting in hypohidrosis or anhidrosis), the hair (wispy), the teeth (fewer in number and conical in shape),⁵ and the nails.⁶ According to Siegel and Potsic, some mesodermal derivatives are also dysplastic in most cases.⁷ Half of these abnormalities are genetically related⁸ and ectodermal dysplasia has classically been considered as an X linked or autosomal recessive condition, which implies that only men can present the complete pattern of the syndrome whereas women can be affected by some of the symptoms only.⁹ However, several females with full blown hypohidrotic ectodermal dysplasia have been reported, which suggests that autosomal recessive transmission may occur.¹⁰ Some authors suggest that ectodermal dysplasia may be an autoimmune disease.¹¹

It is known that a number of entities (estimates run to as high as 117 separate syndromes) make up the ectodermal dysplasia group.¹² Two major forms are recognised: the hidrotic type and the more common anhidrotic or hypohidrotic type.⁷ In order to be as simple as possible, this review of the literature mainly focuses on anhidrotic and hypohidrotic ectodermal dysplasia which are characterised by a triad of partial or complete absence of eccrine sweat glands, sparse hair growth, and deficient teeth.⁷ These dysplasias may be associated with a large number of other abnormalities.⁵

Clinical presentation

The facial features of individuals suffering from ectodermal dysplasia are consistently similar: they usually exhibit fine blond and scanty hair.¹³ Eyelashes and eyebrows are reduced in number or absent. The skin is fine and smooth with deficient sweat and sebaceous glands, and eczematoid and periorbital pigmentation can be seen. Hair follicles are often defective or absent. The nasal bridge is usually depressed with resultant saddle nose configuration. The tenacious consistency of nasal secretions promotes mucous stasis with secondary infection of the nose and paranasal sinuses.¹⁴ Low set and pointed ears may be observed.¹⁵ Chronic cerumen impaction has been reported as a possible manifestation of the disease.^{7 14} External and medium otitis

are common. Lips are protruding and everted.¹⁴ There are various types of ocular manifestations principally involving ectodermal structures such as the cornea, lacrymal systems, lens, and retina.¹¹ These manifestations can result as corneal dysplasia and cataracts.

Intraoral examination can reveal hypodontia, with teeth usually peg or cone shaped. Anodontia is also a possible problem. In certain cases, some authors rely on the size and the number of teeth to establish a diagnosis.¹⁶ Dental deficiencies compromise lip support and are responsible for a decreased lower facial height. Panoramic radiography can be a determinant adjunct in the diagnosis of ectodermal dysplasia. Congenital absence of major salivary glands has been reported¹⁷ resulting in xerostomia, which in turn is reflected by an increased rate of dental decay.

Also found in ectodermal dysplasia patients is hypoplasia or absence of the mucous glands lining the upper aerodigestive tract. It results in chronic upper respiratory tract infections, otitis, dysphagia, hoarseness, bronchitis, and sometimes haemoptysis.⁷ According to Clarke, patients with ectodermal dysplasia are also found to have statistically significant increased prevalence of extrinsic bronchial asthma and allergic rhinitis.¹⁸

Commonly, the skin will present eczematoid changes with reduced hair follicles and sweat and sebaceous glands. Because of a deficient number of sweat glands, patients are unable to perspire and consequently suffer from hyperthermia. This is a potential cause of death and may be responsible for permanent brain damage resulting in the mental retardation occasionally complicating this condition.¹⁹ For that reason, early diagnosis is very important. Despite the profound alteration of hair follicles, the beard is usually well preserved. In some cases there is also nail dystrophy, hyperkeratosis of volar skin, cleft lip and palate, genital anomalies, paradoxical hyperplasia of sebaceous glands in some areas, hypoplastic or absent mammary glands, and a personal or familial history of atopy.^{20 21}

Diagnosis

Even though the diagnosis of ectodermal dysplasia can be considered when unexplained episodes of hyperpyrexia occur, other symptoms can mislead the clinician in establishing the right diagnosis. Cystic fibrosis is occasionally suspected on the basis of respiratory infections.²² Combined with hypothyroidism,²³ the diagnosis can then again be difficult to achieve. Moreover, patients with idiopathic hypoparathyroidism frequently develop ectodermal

disease.²⁴ Martini *et al* report on two young girls in which ozena was the presenting symptom of hypohidrotic ectodermal dysplasia.¹⁰

Because of the possible severity of the disease, early diagnosis is critical. Many diagnostic techniques are available, most of which will be now summarised. Diagnosis in most cases must rely on the determination of presence or absence of functioning sweat glands. Such glands are most numerous on the palms and soles. Therefore, a diagnosis of anhidrotic ectodermal dysplasia may most reliably be made on the basis of determination of number and maturity of glands at those sites.²⁵ Antenatal diagnosis techniques, although more invasive, allow the possibility of therapeutic abortion.

Very helpful for calculating the risk of transmission in affected individuals and females with normal phenotypes is molecular analysis. It can also be applied to chorionic villi sampling in order to achieve antenatal diagnosis in high risk male fetuses. Gene mapping is the best way to confirm antenatal diagnosis of ectodermal dysplasia and skin biopsies under fetoscopy are no longer indicated.²⁶ Clinicians must also be sure they are dealing with the X linked form of hypohidrotic (anhidrotic) form of ectodermal dysplasia.²⁷

Skin biopsy is also no longer indicated to confirm postnatal diagnosis in child or adult. Direct visualisation of the sweat pores with an ophthalmoscope is a safer and non-invasive technique and had replaced older methods such as impression and staining of the skin.²⁸

Finally, perspiration rate can be evaluated by an ionisation method which determines chloride concentration in sweat. It is reported to be a delicate technique which has its limitations and therefore can give equivocal results.²⁹

Management

Because manifestations of ectodermal dysplasia are related to pathological defects that cannot be corrected, alternative measures must be utilised to minimise symptoms.

Myers,¹⁴ as well as Siegel and Potsic,⁷ extensively review the otolaryngological management of patients with ectodermal dysplasia. For that reason, our review of the ear, nose, and throat region will mostly rely on these two articles.

In order to minimise cerumen impaction, routine visits for cerumen disimpaction should be made. The use of lubricating drops (glycerin, mineral oil) on a daily basis as well as peroxide solution weekly is suggested. The treatment of serous otitis media is usually surgical because of the unremitting nature of the disease.

Removal of nasal crusts is often made under general anaesthesia. If possible, gentle mechanical removal can be done by the patient. Alternating irrigation with both saline and gentamicin drops helps in decreasing odours coming from the nose. The use of domestic humidifiers is recommended. Saline nose drops should be used as often as necessary whereas decongestant sprays are not a suitable method of humidifying the nose. In cases of upper respiratory tract infection, these sprays are indicated for temporary symptomatic relief. Appearance can be improved by rhinoplasty.³⁰

Chronic laryngitis caused by inadequate lubrication of the vocal folds may result in hoarseness. Proper humidification should help solving the problem.

The decreased salivary flow observed with some patients can result in difficulties during both mastication and deglutition. The consumption of large quantities of fluid during meals is recommended. For maximum patient comfort, the use of sialogogues between meals can be considered.

In order to optimise the masticatory function and aesthetics, partial or complete dentures should be made as

soon as possible.¹⁸ Osseointegrated implants and implant fixed prosthesis have been successfully used on an adult patient.¹³

Conclusion

Ectodermal dysplasia is a rare but serious disease that has to be diagnosed as soon as possible because of the important problems it can cause. Signs and symptoms occurring in the oral cavity can be very helpful in providing the clinician a provision diagnosis. Even though treatments remain palliative adjuncts, their use is necessary in order to obtain symptom reduction.

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