Hereditary Ectodermal Dysplasia - A Case Report

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Article ID: WMC001711
Article Type: Case Report
Article URL: http://www.webmedcentral.com/article_view/1711
Subject Categories: DENTISTRY
Keywords: Hypodontia, Anodontia, Anhidrosis.
How to cite the article: Babu SG, Castelino RL, Shetty SR, Rao KA. Hereditary Ectodermal Dysplasia - A Case Report. WebmedCentral DENTISTRY 2011;2(3):WMC001711
Source(s) of Funding:
Nil
Competing Interests:
Nil
WebmedCentral Peer Reviewed: Yes
Hereditary Ectodermal Dysplasia - A Case Report

Author(s): Babu SG, Castelino RL, Shetty SR, Rao KA

Abstract

Ectodermal dysplasia is an extremely rare genetic disorder characterized by faulty development of ectodermal structures. The tissues in which the primary defects occur are the skin, hair, nails, exocrine glands, and teeth. It usually affects the males and females are the carriers. The clinical features include sparse hair, abnormal or missing teeth, and inability to sweat due to lack of sweat glands. One such case report of ectodermal dysplasia is presented here.

Introduction

Ectodermal dysplasias are a heterogeneous group of disorders characterized by developmental dystrophies of ectodermal structures. It is an X linked recessive disorder. It is characterized by the triad of signs which comprises of sparse hair (atrichosis or hypotrichosis), abnormal or missing teeth (anodontia or hypodontia) and inability to sweat due to lack of sweat glands (anhidrosis or hypohidrosis). The etiology of ectodermal dysplasia appears to be genetic in nature [1].

Case Report(s)

A 32-year-old male patient reported to our institution with the complaint of multiple missing teeth since childhood. The patient also gave a history of delay in the eruption of deciduous and permanent teeth, intolerance to heat and reportedly less sweat production. There was no history of consanguineous marriage between the parents. On extra oral examination, the patient had dry skin with periorcular area being hyperpigmented and wrinkled with sparse hair on the body and scalp. Hair present were fine in texture & lighter in color. Prominent supraorbital ridges, small and outwardly placed ears and flattened nasal bridge was also present. Both upper and lower eyelids showed sparse eyelashes [Figure 1 and 2]. The skin was warm and dry.

Intra oral examination revealed multiple missing teeth in the maxillary and mandibular arches [Figure 3]. Salivary Flow rate was estimated at 0.1 ml in 5 min. The similar findings were present in the maternal uncle, as revealed by the patient. Based on these findings a diagnosis of hereditary ectodermal dysplasia was made. An orthopantomograph was made which revealed multiple missing teeth [Figure 4]. The teeth with bad prognosis were extracted and the replacement of missing teeth was done using a fixed prosthesis in the maxillary and mandibular arches [Figure 5].

Discussion

The Ectodermal dysplasias are a group of inherited disorder that share in common developmental defects involving at least two of the major structure classically hold to derive from the embryogenic ectoderms hair, teeth, nails, sweat glands [2]. The incidence of ectodermal dysplasia in males is estimated at 1 in 100,000 births [3]. Thurman first described this entity in 1848[4]. More than 192 distinct disorders have been described till date. Freire Maia and Pinheiro published an exhaustive review and classification system for these disorders using a numeric system of 1 (hair), 2 (teeth), 3 (nail), 4 (sweat glands) for characterization [5]. The complete syndrome does not occur in females but females may show dental defects, sparse hair, reduced sweating and dermatoglyphic abnormalities [2]. The major concern seen in these patients is the lack of teeth and the special appearance, as seen in our case [1]. The most characteristic findings is the reduced number and abnormal shape of teeth. The delay in eruption of teeth is often the first step in the diagnosis. The men have an easily recognizable facies, also referred to as an old man facies. Some infants have a premature look with scaling of the skin. This can also form a clue to the diagnosis [1]. The extra oral features seen in this disorder are frontal bossing with the forehead appearing square in shape, prominent supraorbital ridge, depressed nasal bridge (saddle nose) as seen in our case. The other features include midface hypoplasia, pointed chin and protruberent and everted lips, however these features were not seen in our case [6]. Abnormalities of skin, nails and teeth are also noted, which was also seen in our case [1]. Prenatal diagnosis of ectodermal dysplasia has occasionally been reported which has been diagnosed by foetal skin biopsy, obtained by fetoscopy by 20 weeks gestation [7]. The characteristic facies is pathognomonic but may not be recognized in infancy. In partial forms, the pointed
conical teeth provide the most valuable indication and should suggest the need for sweat test and a skin biopsy [2]. Both autosomal dominant and recessive mode of inheritance has been described. Another variety of hydrotic ectodermal dysplasia called as Clouston syndrome is inherited in an autosomal dominant manner, was described by Clouston in 1929 and Lowrey et al in 1966, which is found in Canadian families of French descent. This syndrome usually spares the sweat glands [8]. The treatment usually comprises of complete restoration of function and aesthetics normalise the vertical dimension and provide adequate support to the facial soft tissues.

Conclusion

Ectodermal dysplasia is a rare genetic disorder with involvement of various tissues in the body. A careful and a thorough examination will lead to an accurate diagnosis. Restoration of normal function should be the main concern in these patients.

References

Illustrations

Illustration 1

Extra oral photograph showing sparse hair on the face and scalp.

Illustration 2

Sparse body hair
Illustration 3

Intra oral photograph showing multiple missing teeth

Illustration 4

Orthopantomograph showing multiple missing teeth
Illustration 5

Post treatment photograph of the patient
Reviews

Review 1

Review Title: Hereditary Ectodermal Dysplasia

Posted by Dr. William J Maloney on 27 Sep 2012 08:36:10 PM GMT

What are the main claims of the paper and how important are they?:
The main purpose of this article is to describe a case of hereditary ectodermal dysplasia.

Yes

If a protocol is provided, for example for a randomized controlled trial, are there any important deviations from it? If so, have the authors explained adequately why the deviations occurred?

Yes

Rating: 8

Comment:
Ectodermal dysplasias are a group of disorders which are characterized by developmental dystrophies. It is an x-linked recessive disorder. The etiology of ectodermal dysplasia is genetic. The skin, teeth, hair, nails, and exocrine glands are affected.

The authors conclude that a careful and thorough examination will lead to an accurate diagnosis and that the restoration of normal function should be the main concern in these patients.

Competing interests: None

Invited by the author to make a review on this article?: No

Have you previously published on this or a similar topic?: No

Experience and credentials in the specific area of science:
Clinical associate professor

How to cite: Maloney W.Hereditary Ectodermal Dysplasia[Review of the article 'Hereditary Ectodermal Dysplasia - A Case Report ' by ].WebmedCentral 1970;3(9):WMCRW002258
Review 2

Review Title: Hereditary Ectodermal Dysplasia - Case Report

Posted by Dr. Nandita Shenoy on 25 Nov 2011 05:39:37 AM GMT

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Rating: 5

Comment:

- Ectodermal dysplasia is not an extremely rare genetic disorder as stated in the manuscript
- "Ectodermal dysplasia is an X linked recessive disorder" which is not true. Ectodermal dysplasias are can be autosomal dominant, autosomal recessive and X-liked disorders.
- This condition can affect males and females equally.
- Method of estimation of salivary flow not mentioned
- Please check English grammar and style

- Discussion should have been a little longer.
- Genetic levels and grades are not discussed in detail.
- Rehabilitation and prosthetic management could have been discussed
- Overall not very well documented and not a rare condition to report.

Competing interests: No

Invited by the author to make a review on this article?: No

Have you previously published on this or a similar topic?: No

Experience and credentials in the specific area of science:
This is the specialty i deal with every day.

How to cite: Shenoy N. Hereditary Ectodermal Dysplasia - Case Report [Review of the article 'Hereditary Ectodermal Dysplasia - A Case Report' by ]. WebmedCentral 1970;2(11):WMCRW001157
Review 3

Review Title: Hereditary Ectodermal Dysplasia - A Case Report

Posted by Dr. Balendra Pratap P Singh on 17 Oct 2011 06:54:14 AM GMT

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Rating: 4

Comment:
well presented the diagnosis part but management of such case need to be presented in detail like is it tooth supported fixed prosthesis or implant supported prosthesis? what about the follow-up problems? what were the post-operative instructions given to the patient?

Competing interests: no

Invited by the author to make a review on this article? : No

Have you previously published on this or a similar topic?: No

Experience and credentials in the specific area of science:
follow-up of three years of such hereditary ectodermal dysplasia cases.

How to cite: Singh B. Hereditary Ectodermal Dysplasia - A Case Report [Review of the article 'Hereditary Ectodermal Dysplasia - A Case Report ' by ]. WebmedCentral 1970;2(10):WMCRW001017
Review 4

Review Title: hereditary ectodermal dysplasia- a case report

Posted by Dr. claudia p dellavia on 12 Apr 2011 04:38:58 PM GMT

1. Is the subject of the article within the scope of the subject category? Yes
2. Are the interpretations / conclusions sound and justified by the data? No
3. Is this a new and original contribution? No
4. Does this paper exemplify an awareness of other research on the topic? No
5. Are structure and length satisfactory? No
6. Can you suggest brief additions or amendments or an introductory statement that will increase the value of this paper for an international audience? No
7. Can you suggest any reductions in the paper, or deletions of parts? No
8. Is the quality of the diction satisfactory? No
9. Are the illustrations and tables necessary and acceptable? No
10. Are the references adequate and are they all necessary? No
11. Are the keywords and abstract or summary informative? No

Rating: 2

Comment:
abstract is too general

update references

check english grammar and style

expand discussion

add some other images

no significant contribution to this specific research field is provided

there is confusion concerning the classification of Ectodermal dysplasias: please specify that the present patient was afflicted with Hypohidrotic form

no multidisciplinary treatment was done: no aesthetich concerns, speech problems, detailed facial features... were analysed

Competing interests: no

Invited by the author to make a review on this article? : No
Have you previously published on this or a similar topic?: Yes


Experience and credentials in the specific area of science:

anthropometric analysis of facial features in subjects with ectodermal dysplasia: cross-sectional and longitudinal studies

How to cite: dellavia c.hereditary ectodermal dysplasia- a case report[Review of the article 'Hereditary Ectodermal Dysplasia - A Case Report ' by ].WebmedCentral 1970;2(4):WMCRW00667
Review 5

Review Title: Ectodermal dysplasia

Posted by Dr. Mahfujul H Khan on 09 Apr 2011 05:21:46 AM GMT

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Rating: 5

Comment:
It may be accepted for publication

Competing interests: no

Invited by the author to make a review on this article?: No

Have you previously published on this or a similar topic?: Yes


Experience and credentials in the specific area of science: 20 years

How to cite: Khan M. Ectodermal dysplasia [Review of the article 'Hereditary Ectodermal Dysplasia - A Case Report ' by ].WebmedCentral 1970;2(4):WMCRW00650
Review 6

Review Title: good article

Posted by Dr. Anusha R L on 03 Apr 2011 03:51:45 PM GMT

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Rating: 6

Comment:
The article is substantially good

Competing interests: No

Invited by the author to make a review on this article?: Yes

Have you previously published on this or a similar topic?: No

Experience and credentials in the specific area of science:
1 year

How to cite: R L A.good article[Review of the article 'Hereditary Ectodermal Dysplasia - A Case Report ' by ].WebmedCentral 1970;2(4):WMCRW00638
Review 7

Review Title: Hereditary Ectodermal Dysplasia - A Case Report

Posted by Dr. Prasanna K Rao on 28 Mar 2011 12:07:31 PM GMT

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Rating: 7

Comment:
Overall the article is fairly good.

Competing interests: Nil

Invited by the author to make a review on this article?: Yes

Have you previously published on this or a similar topic?: Yes

References: KARNATAKA state DENTAL journal 2005 May issue.

Experience and credentials in the specific area of science:
6 years of teaching experience in Oral Medicine and Radiology.

Review Title: Hereditary Ectodermal Dysplasia - A Case Report

Posted by Prof. Mostafa I Mostafa on 16 Mar 2011 10:13:48 AM GMT

1. Is the subject of the article within the scope of the subject category? Yes
2. Are the interpretations / conclusions sound and justified by the data? No
3. Is this a new and original contribution? No
4. Does this paper exemplify an awareness of other research on the topic? No
5. Are structure and length satisfactory? Yes
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7. Can you suggest any reductions in the paper, or deletions of parts? Yes
8. Is the quality of the diction satisfactory? Yes
9. Are the illustrations and tables necessary and acceptable? Yes
10. Are the references adequate and are they all necessary? No
11. Are the keywords and abstract or summary informative? No

Rating: 2

Comment:
Dear Webmed Central team, thank you for your invitation to review the article entitled "Hereditary Ectodermal Dysplasia - A Case Report" by Babu S et al. My comments are as follows:

- Regarding abstract, authors mentioned that "Ectodermal dysplasia is an extremely rare genetic disorder" which is not true. It is a rare disorder or a common genetic disorder. Authors also mentioned that "It usually affects the males and females are the carriers". This is true only in the X-linked type while the other 2 types (autosomal dominant and autosomal recessive) affect both males and females equally.

- Regarding introduction, authors mentioned that "Ectodermal dysplasia is an X linked recessive disorder" which is not true. Ectodermal dysplasias are characterized by genetic heterogeneity i.e. transmitted as autosomal dominant, autosomal recessive and X-linked disorders.

- OMIM (Online Mendelian Inheritance in Man) number should be mentioned.

- Regarding case report, I estimated that the present case is an X linked type. The pedigree (family tree) should be taken. Mother is a carrier of the affected gene and should be examined because partial manifestations as missing upper lateral incisors could be reported.

- Regarding discussion, authors ignored to discuss the suggested treatment regarding periodontal condition and previously published treatment modalities.
Finally I have a negative decision. The article reported a well-known and well-documented condition and did not describe the management in detail without a long-term follow-up.

Thank you.

Competing interests: Not

Invited by the author to make a review on this article? : No

Have you previously published on this or a similar topic?: Yes

References:

Experience and credentials in the specific area of science:

Yes

How to cite: Mostafa M.Hereditary Ectodermal Dysplasia - A Case Report[Review of the article 'Hereditary Ectodermal Dysplasia - A Case Report ' by ].WebmedCentral 1970;2(3):WMCRW00590
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