Hypohidrotic ectodermal dysplasia: dental, clinical, genetic and dermatoglyphic findings of three cases

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Patients with hypohidrotic ectodermal dysplasia (HED) are characterized by the clinical manifestations of hypodontia, hypohidrosis, hypotrichosis and a highly characteristic facial physiognomy. This disorder is inherited as an X- linked trait. This report presents three cases with HED in which the clinical evaluation (intraoral and radiological), genetic findings and SEM examination of hair. Boys 6 to 14 year old and a 11 year old girl were referred to the Marmara University, Faculty of Dentistry, complaining of oligodontia in the maxillary and mandibular arches and delay in eruption of other teeth. Peg-shaped teeth have been observed. The dermatoglyphs of the patients were striking. SEM exmimation of hair demostrated a distinctly abnormal longitudinal groving along the entire length of each hair and a desquamation of the surface cuticles. The treatment was planned in a multidisciplinary odontological group involving pediatric dentistry, orthodontics, prosthodontics and oral surgery and maxillofacial radiology of future dental habilitation. A specially designed overdenture, a removable prosthesis and osseointegrated implants were constructed. Periodic recall visits were advised, to monitor the dentures and implants during periods of growth and development, and eruption of the permanent teeth.
J Clin Pediatr Dent 26(1): 5-12, 2001

INTRODUCTION

Chilдren with ectodermal dysplasia present diagnostic and treatment challenges because of the many oral manifestations. Ectodermal dysplasias consists of clinical and genetic heterogeneous groups of disorders, characterized by either absence incomplate or delayed development of one or more of the appendages derived from epidermal tissue (hair, sweat gland and nails) or of oral ectodermal origin during embryogenesis.

Patient with hypohidrotic ectodermal dysplasia (HED) are characterized by the clinical manifestations of hypodontia, hypohidrosis, hypotrichosis and a highly characteristic facial physiognomy. This disorder is inherited as an X- linked trait.

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CASE REPORTS

CASE 1

A eight year old female (Figure 1) (III-10) with characteristic apparence was admitted to the Pediatric Dentistry Department with main complaints of chewing difficulties and esthetic problems. The parents (II-1 and II-7) were first cousins. The patient was the second child of her family (Figure 2). Her body was devoid of lamigo hair. She was blonde. Her scalp hair was stiff and short. SEM view of the hair was shown desquaqmation of the surface cuticles (Figure 3). The eye lashes and eye brows were scanty. She had frontal bossing and depressed nasal bridge. There were linear wrinkles and pigmentation about the eyes. Her lips were protuberant due to absence and maldeveloped teeth (Figure 4). The skin was soft, thin and dry. She had inability to sweat. The nails were normal. Her pharyngeal and laryngeal mucosa were atrophic resulting in dysphonia. She had breathy voice due to atrophy of the oral mucosa. The nipples were rudimantary. Cephalometric study showed a small lower facial height and depth, small palatal and cranial basewidths. Small valar bones, small calvarial height. Intraoral views demonstrates cone shaped teeth and oligodontia (Figures 5, 6).

Dermatoglyphic findings of the patient and her parents are shown in Table 1 and Figure 7. There were eight arches on the finger-tips of the patient. (III-10) The total finger ridge-count (TRC) was lower than

Figure 1. Facial view of the patient 1.

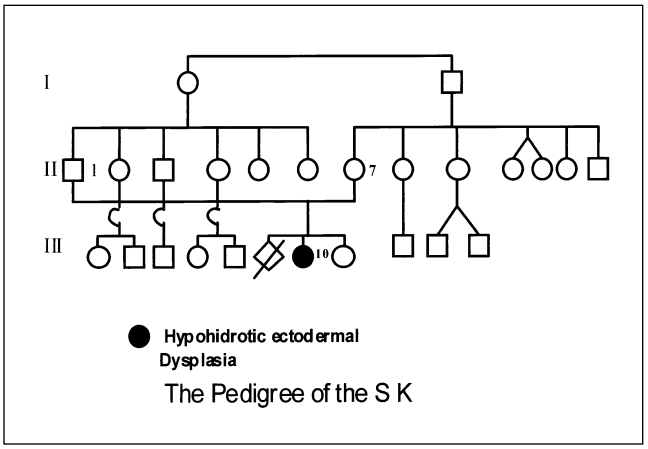


Figure 2. The pedigree of the patient 1.

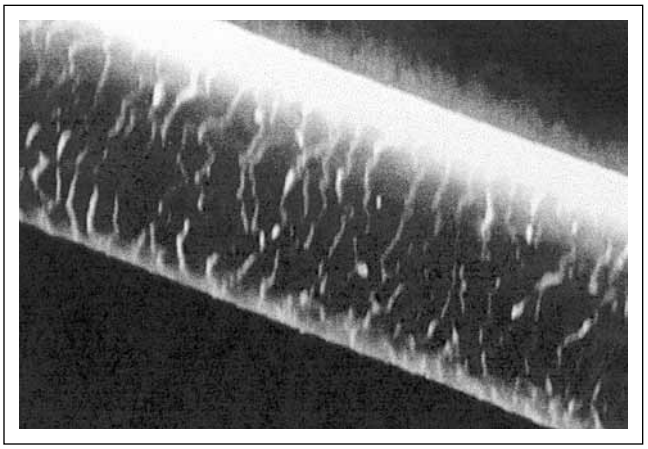


Figure 3. SEM view of the hair.

those of the female cases. The mean for TRC's of the female control cases was x=137.76 ± 2.22 (n=447). The summed palmar a-b ridge-counts of the control cases was x=80.19 ± 0.54 (n=500). There were hypotenar ulnar loops (H'^{s}) associated distally displaced axial triadii on the palms. The A-line was terminating on the radial border of the left hand. The pattern intensities on both palms and right sole were high. There were 6 arches on the finger-tips of the mother (II-7). There was an arch on the right index finger of the father (II-1). The mother had unusually rare pattern, which is called radial mutant loop on her right thumb. This pattern type was not observed on the right fingers of 500 control cases. Both the father and the mother had less TRC's on the fingers. The mean for TRC's of the male control cases was x=151.71 ± 2.13 (n=446). The sweat pore counts in 1 cm^2 on the palms of the patient, her father and mother were 27, 76 and 84 respectively The means for the sweat pore number in 1 cm^2 on the palms of the male and female control cases were x=126.33 ± 0.99 (n=160) and x=151.44 ± 0.83 (n=140), respectively.

After the exfoliation of the primary incisors a removable partial dentures is preferred. (Figures 8, 9)

CASE 2

A four year old male (Figure 10) (III-11) with characteristic apparence admitted to the Department of Pedodontics with main complaints of dental caries, chewing difficulties and esthetic problems. The skin was soft, thin and dry. She could not tolerate heat well. The parents (II-5 and II-14) were first cousins. The patient was second child of the family (Figure 11). His scalp hair

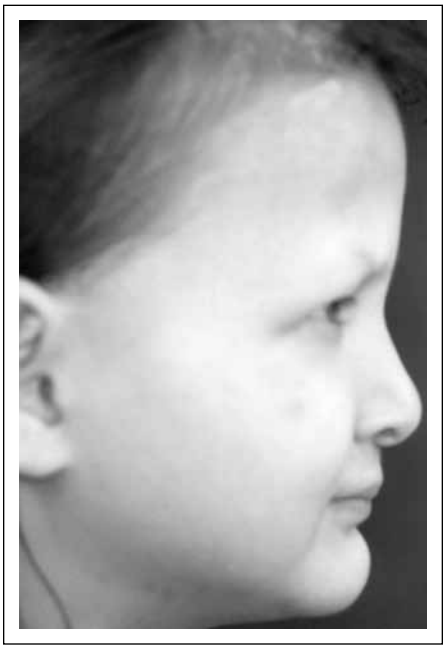
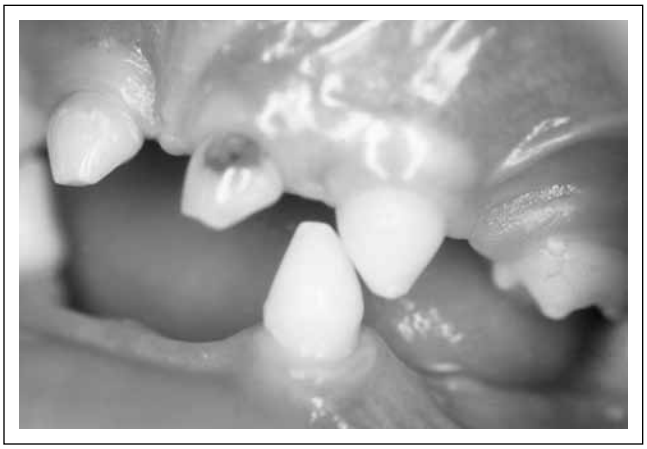


Figure 4. Profile of the patient 1.



Figures 5, 6. Intraoral view of the patient 1.

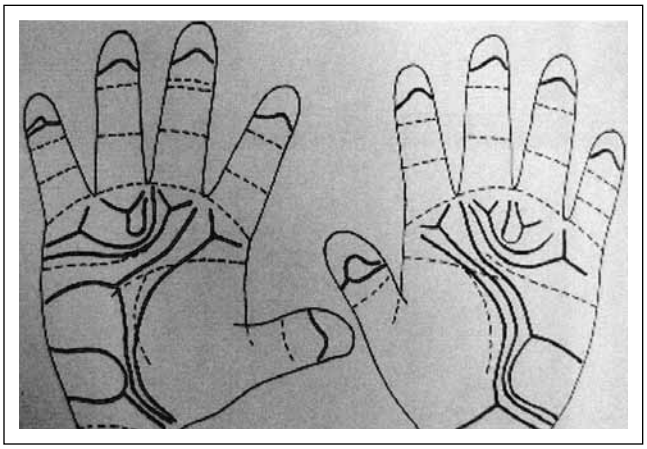
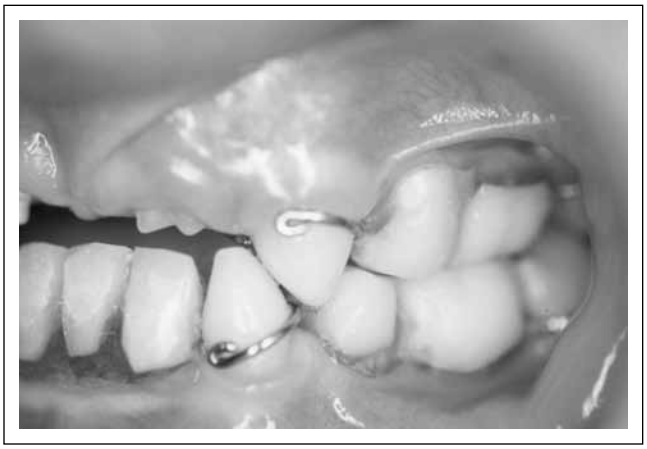
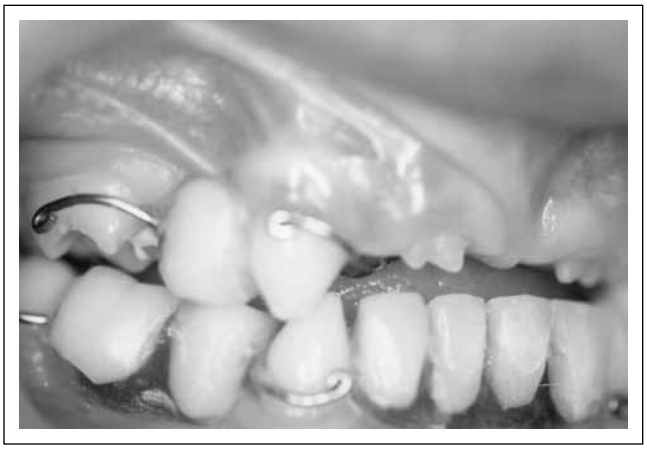


Figure 7. The dermatoglph of the patient 1.



Figures 8. Intraoral view showing the removable partial dentures.



Figures 9. Intraoral view showing the removable partial dentures.

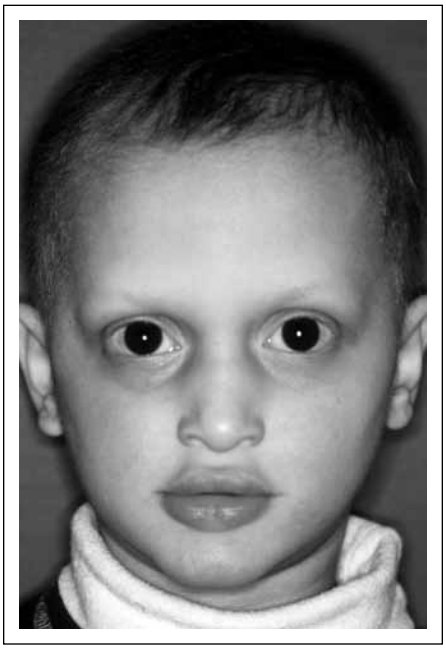


Figure 10. Facial view of the patient 2.

was blond, stiff and short SEM view of the hair demonstrated a desquaqmation of the surface cuticles (Figure 12). The eyelashes and eyebrows were loosely woven. He had frontal bossing and depressed nasal bridge. His lips were protuberant. (Figure 13). The incisor teeth and canines were conical in apperance. Intraoral views

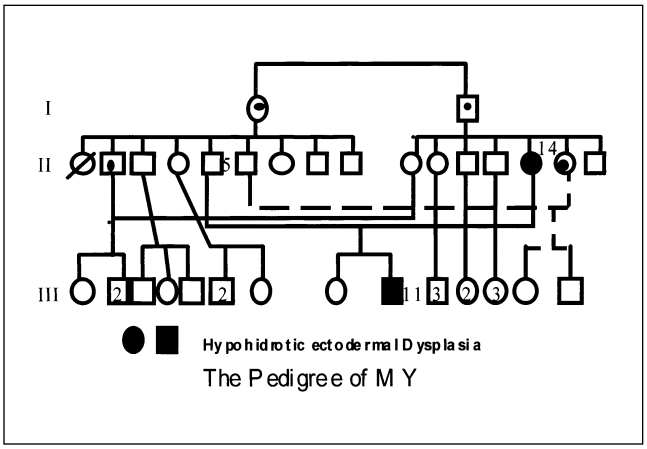


Figure 11. The pedigree of the patient 2.

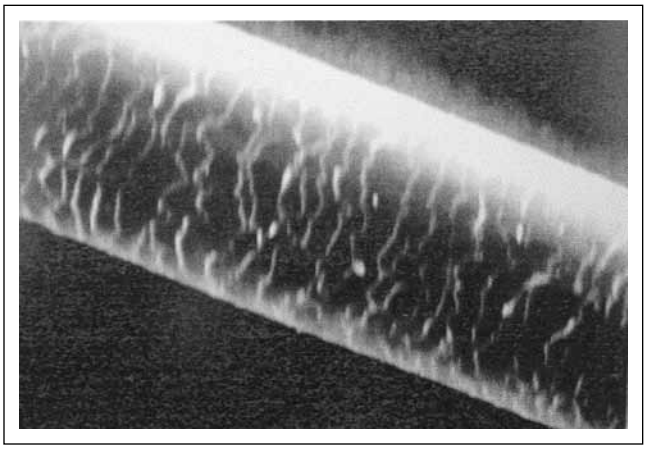


Figure 12. SEM view of the hair.

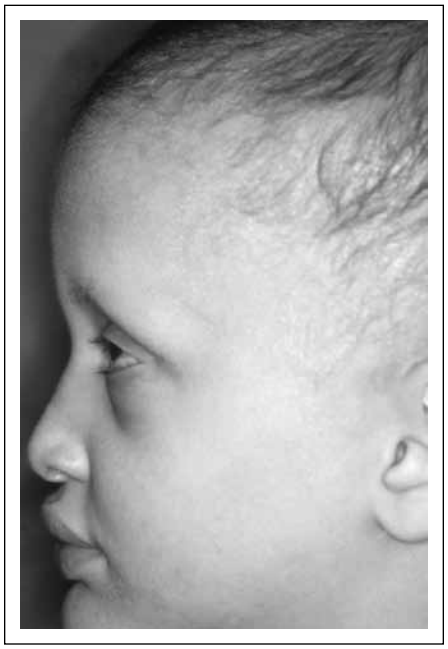


Figure 13. Profile of the patient 2.

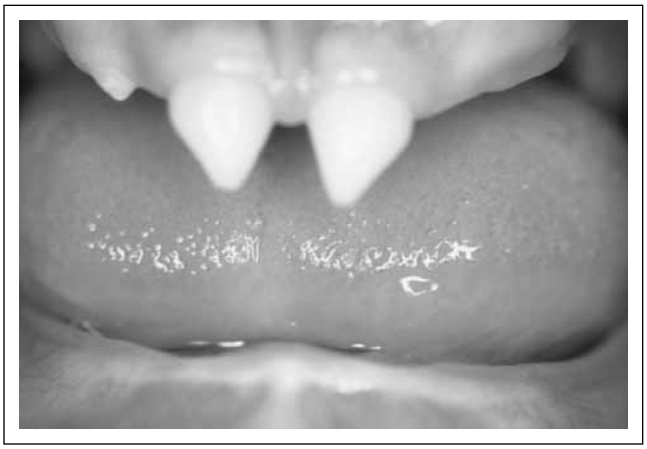


Figure 14. Intraoral view of the patient 2.

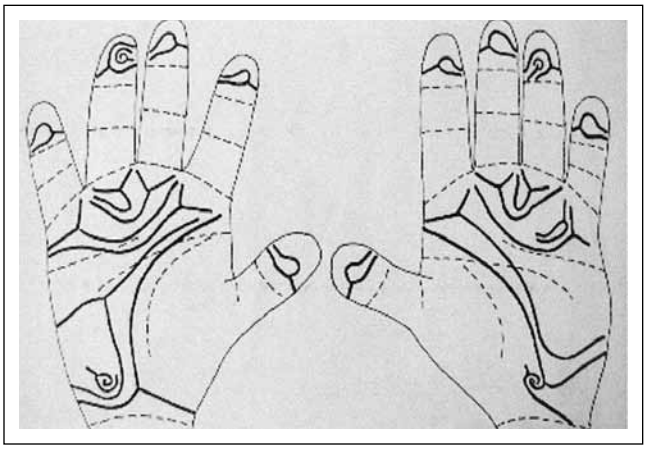


Figure 15. The dermatoglph of the patient 2.

demonstrates anadontia at the mandible and oligodontia at the maxilla. The incisor teeth and canines were conical in apperance (Figure 14).

The dermatoglyphic findings of patient 2 and his parents are shown in Table 1 and Figure 15. The TRC was lower. The mean for TRC's of the male control cases is x=150.79 ± 2.84(n=25). There was a distally displaced axial triradius and were H and $\hat{H}$ loops on the left hypothenar area. There were H and $\hat{H}$ loops, one originating from a radiant of axial triradius on the right palm. The pattern intensities on both palms lower. The ridges were distorted mainly on the left sole. The sweat pore number was reduced. The dermatoglyphics of the father (II-5) was not characteristic. The mother (II-14) had distally displaced axial triadii on her both palms.

The maxilla received a fixed denture and the mandible received an osteointegrated implant supported denture. (Figures 16 to 18).

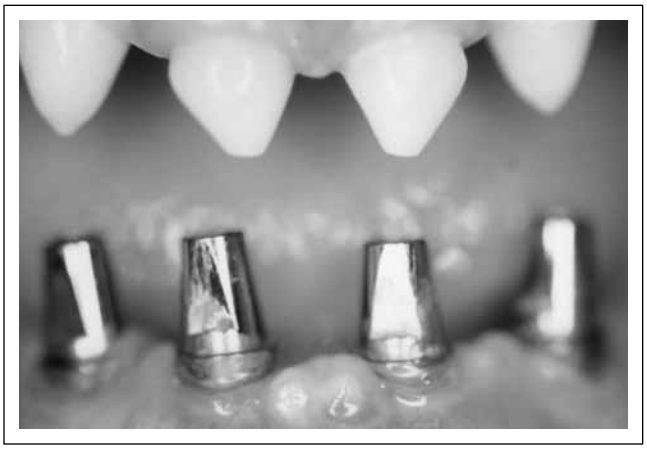


Figure 16. Intraoral view showing osseointegration of the implants.

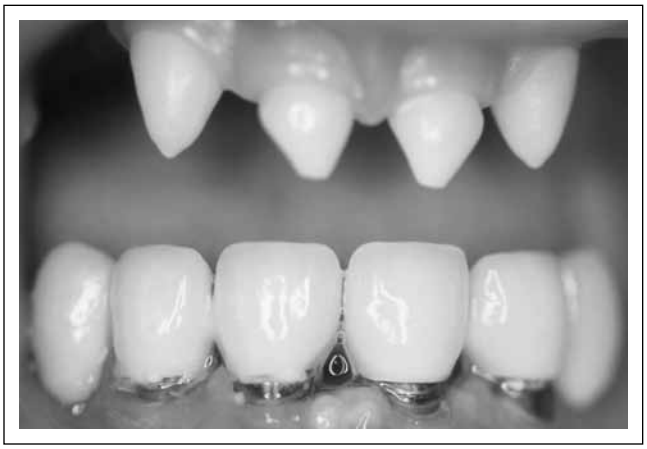


Figure 17. Intraoral view showing upper prosthesis.

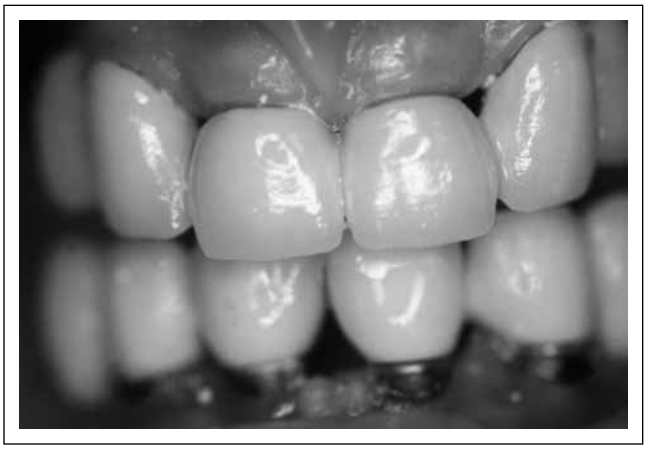


Figure 18. Intraoral view showing lower and upper prosthesis.

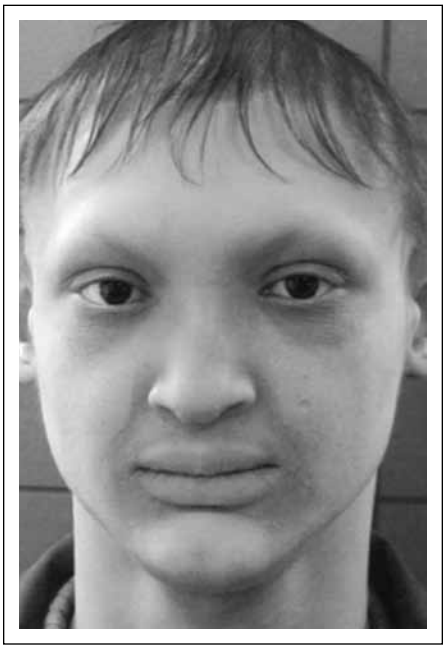


Figure 19. Facial view of the patient 3.

CASE 3

An eleven old male (Figure 19) (IV-8) came to the Department of Pedodontics with main complaints of chewing difficulties and esthetic problems. The parents (III-2 andIII-5) were second cousins. The patient was the third child of the family. (Figure 20). His hair was sparse and blond. SEM view of the hair demonstrated a desquaqmation of the surface cuticles (Figure 21). He had eczema and hypohidration. The lips were protuberant due to absence of the frontal teeth and conical shaped incisors. He had frontal bossing and depressed nasal bridge (Figure 22). Panoramic radiograph indicates absence of primary or permanent teeth (Figure 23). The intra-oral views demonstrates typical conical shaped incisors and molar (Figure 24). The dermatoglyphic findings of patient 3 and his parents are shown in Table 1 and Figure 25.

The TRC was lower. There was a H loops associated with intermediately placed axial triradius on his left palm. There were Ĥ loops on the hypo thenar area of the right palm . The ridges containing a few pores were distorted particularly on the left hypo thener area of the left palm. The dermatoglyphics of the parents were not remarkable.

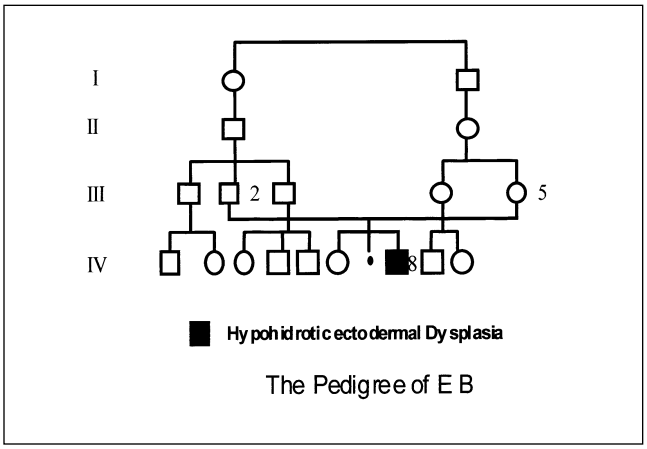


Figure 20. The pedigree of the patient 3.

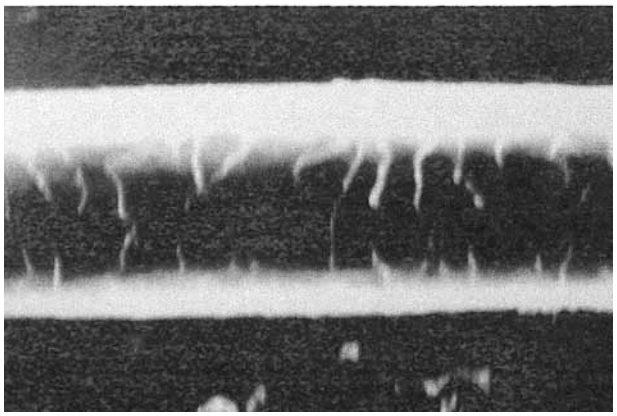


Figure 21. SEM view of the hair 3.

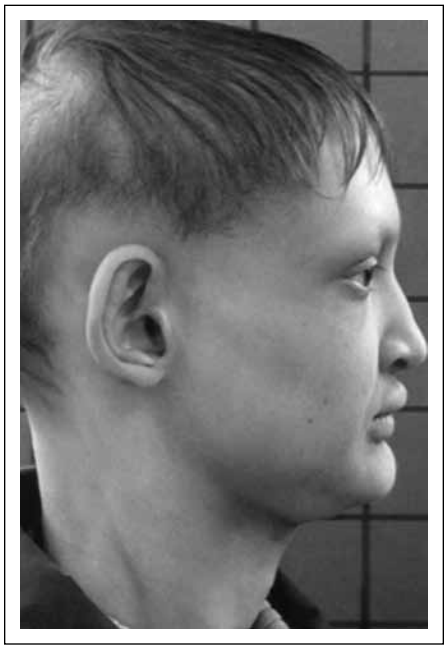


Figure 22. Profile of the patient 3.

Full metal crowns for incisors and overdenture are prepared for the maxilla. Removable partial denture is prepared for the mandibular arch (Figures 26, 27).

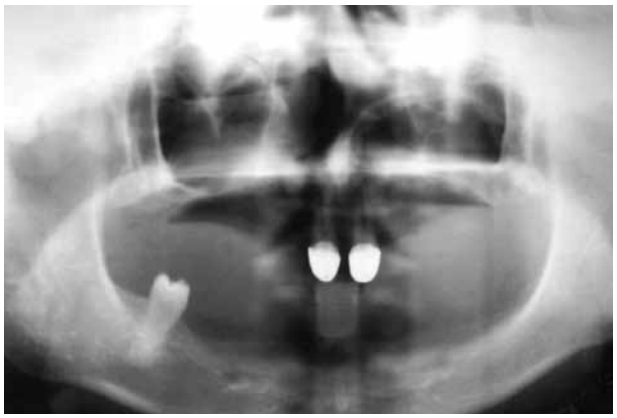


Figure 23. Panoramic radiograph of the patient 3.

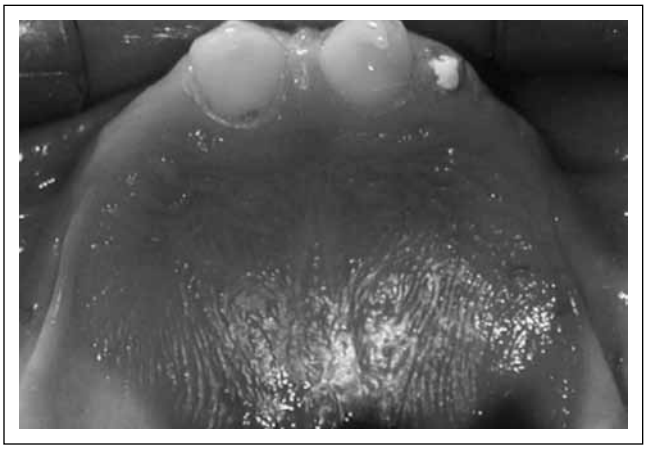


Figure 24. Intraoral view of the patient 3.

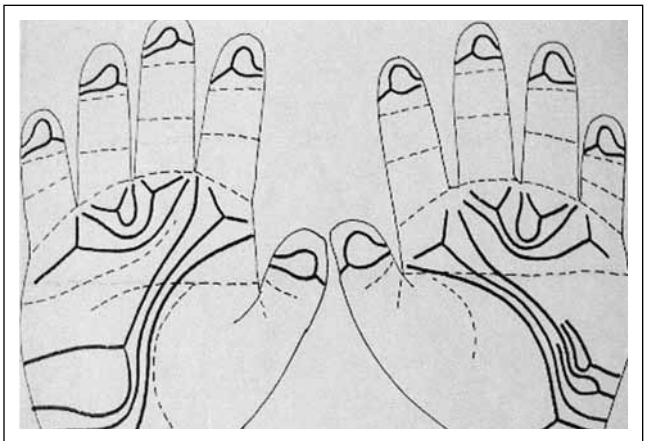


Figure 25. The dermatoglph of the patient 3.

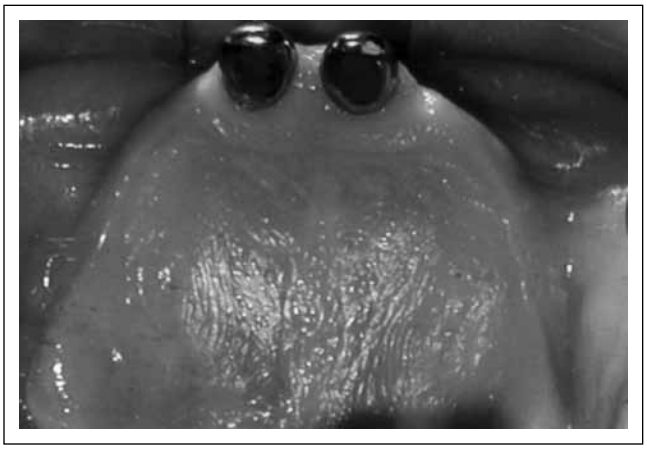


Figure 26. Intraoral view showing upper full crowns.

Table 1. Finger-tip, palmar and plantar configuration types, total finger and summed palmar a-b ridge-counts of the patients and the parents.

		Fingers V IV III II I	TRC	Plantar formulae	Summed palmar a-b ridge-count	Palmar formulae	Sweat pore count
Patient 1 (III-10)	L R	U A A A A A A A A U	11	III H t" t 4 (1) IV H t" t 4 (3)	31	I IV f p^{l} 4 I III IV f p p^{ll} 4	12
The mother of the patient 1 (II-7)	L R	U A A A A U U A A R	45	I^{r} III e t' 4 (5^{l}) III t' 4 (5^{l})	90	I III f p 4 I III f p 4	18
The father of the patient 1 (II-1)	L R	U W U R U U W U A U	125	III^{T} t 4 (4) III $\hat{H}$ t t^{b} 4 (4)	82	I II III III f p z 4 I III III f p z z^{l} 3	17
Patient 2 (III-11)	L R	U W U U U U W U U U	50	IV H $\hat{H}$ t^{ll} t 4 (5^{l}) III IV^{r} H $\hat{H}$ t t 5 (3)	94	4 I 4	12
The mother of the patient 2 (II-14)	L R	U A A U U U U U U U	57	t" 3 (1) III t" 4 (4)	70	I II III f p z 4 I IV f z 3	18
The father of the patient 2 (II-5)	L R	U W U W W U W W W W	101	III t 4 (4) III t 4 (4)	79	I I II e f 4 I f 4	19
Patient 3 (IV-8)	L R	U U U R U U U U U U	48	III $\hat{H}$ t' t 4 (3) III $\hat{H}$ $\hat{H}$ t t^{b} t^{b} 4 (3)	87	I IV e 4 II IV IV f 5	17
The mother of the patient 3 (III-5)	L R	U U U U U U U U R W	115	IV t 4 (3) IV t 4 (3)	80		20
The mother of the patient 3 (III-2)	L R	U U U U U U U U U U	144	IV t 4 (4) IV t 4 (3)	86		22

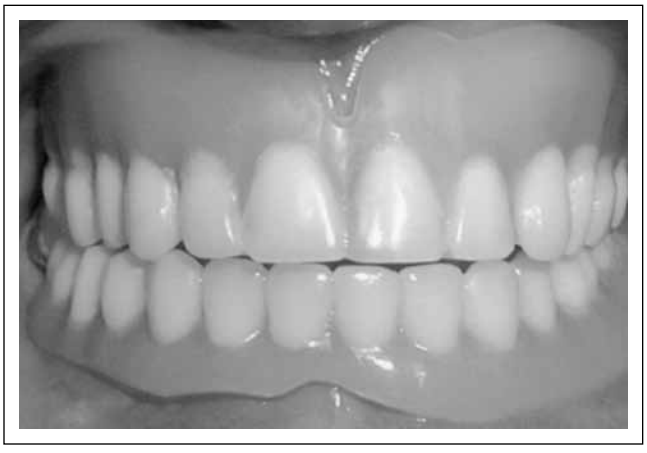


Figure 27. Intraoral view showing total prosthesis.

DISCUSSION

The pedigree investigations of the patients presented here showed that the parents were heterozygous carriers for an autosomal recessive gene, which determined HED. The autosomal recessive inheritance pattern in present patients came from consanguineous marriages observed in the present report. It is well known that the chance of being homozygote affected in this type transmission increased with consanguineous marriages. The frequency of consanguineous marriages in the Turkish population is 29 %. All the present patients were from this type of marriages.

The patient with HED had no sweat-pores. The carrier mothers had fewer sweat-pore counts and no sweat-pores were found in male hemizygotes with the x-linked form. We found diminished sweat-pore counts in both patients with HED and the carrier parents.

Dental findings of the present patients supported the finding of the previously presented ones in the literature. Indeed, congenital hypodontia of maxillary lateral and mandibular primary incisors, and conical crowns of central incisors and maxillary second premo-

lars with supernumerary cusps and taurodontism of maxillary and mandibular second primary molars and diestema between maxillary central incisors are constant findings in HED.

All hair were sparse and blond. SEM view of the hair demonstrated a desquamation of the surface cuticles.

Children and adolescents with ectodermal dysplasia often need extensive and complicated prosthetic treatment due to hypodontia. Functional, esthetic and psychological reasons make important to start such oral habilitation early in life. Most of the reports in the literature concernig treatment are case reports dealing with different types of removable dentures. The development of techniques for osseointegrated implants offers new and better possibilities for the habilitation of the oral conditions of the children. However this habilitation involves a number of problems for example, consideration of growth, tooth development and eruption, type of prosthetics with respect to age, timing of the treatment, etc.

The treatment must be planned in a multidisciplinary odontological group involving pediatric dentistry, orthodontics, prosthodontics and oral surgery and maxillofacial radiology of future dental habilitation.

Periodic recall visits were advised, to monitor the dentures and implants during periods of growth and development, and eruption of the permanent teeth.