

## Palatal Size and Shape in 6-Year Olds Affected by Hypohidrotic Ectodermal Dysplasia

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### ABSTRACT

**Objective:** To analyze the size and shape of the hard tissue palate of Italian subjects with hypohidrotic ectodermal dysplasia (HED).

**Materials and Methods:** The morphology and the dimensions of the hard tissue palate were analyzed in eight 6-year-old boys affected by HED. Four of the boys were completely edentulous and four partially dentate. Palatal landmarks were identified on stone casts and digitized with three-dimensional computerized electromagnetic instrumentation. Palatal length, slope, width, and maximum palatal height in both the sagittal and frontal planes were measured. From the coordinates of palatal landmarks, a mathematical equation of palatal shape was constructed, independent of size. HED palatal data were compared with reference data obtained from 12 healthy boys with a complete deciduous dentition.

**Results:** Palatal length and height in both the sagittal and frontal planes were significantly reduced in HED as compared with control individuals. A less steep (not significant) palatal slope was found in HED than in reference subjects, whereas similar palatal width values were observed. All palatal measurements were larger in partially dentate than in edentulous patients. Both HED and edentulousness influenced palatal shape. The HED boys had a relatively lower palate than the reference boys. In the edentulous HED boys, the hard tissue palate was relatively lower than in partially dentate HED subjects.

**Conclusions:** Palatal size and shape were significantly modified by the presence of hypohidrotic ectodermal dysplasia, and the major alterations were found in edentulous HED subjects.

**KEY WORDS:** Morphometry; Hypohidrotic ectodermal dysplasia; Palate

### INTRODUCTION

Ectodermal dysplasias (ED) represent a large, heterogeneous group of inherited disorders that share primary defects in the development of two or more tissues derived from ectoderm, such as skin, hair, nails, eccrine glands, and teeth. Over 150 types of ED have

been described and classified.<sup>1</sup> The most common syndrome is known as hypohidrotic ectodermal dysplasia (HED), with a frequency of about 1 per 10,000–100,000 live births.<sup>2,3</sup>

Individuals affected by HED present a classical triad of hypotrichosis, hypohidrosis, and hypodontia. Hair is sparse and light-pigmented, and the ability to sweat is greatly reduced. The dental manifestations affect the primary and permanent teeth, with the crowns of the maxillary incisors and canines often being conical or pegged.<sup>4</sup>

Clinical management of oligodontia presents the prosthodontist with peculiar aesthetic and functional needs.<sup>5</sup> Removable prostheses (complete/partial dentures or overdentures, according to the pattern of teeth) are the most frequent treatments during childhood.<sup>3</sup> Regular follow-up is very important in the management of HED patients, and the loss of prosthesis retention, sore points, and occlusal changes caused by erupting teeth or jaw growth should be carefully monitored by the prosthodontist.<sup>5,6</sup>

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**Table 1.** Maxillary Dental Formula in Partially Dentate Patients Affected by HED<sup>a</sup>

HED Subject	17	16	55	54	53	52	51	61	62	63	64	65	26	27
M5	(x)		x		x		x			x				(x)
M6			x		x			x		x		x		
M7			x		x		x	x		x		x		
M8	(x)	x	x			x	x	x				x	x	(x)

<sup>a</sup> HED indicates hypohidrotic ectodermal dysplasia; (x), dental germ present at radiographic examination; and x, erupted tooth.

Moreover, in the craniofacial complex, structures derived from the mesoectodermal layer of the neural crest are quite often involved, producing an abnormal morphology.<sup>7</sup> In HED patients, craniofacial anomalies have been reported in several cephalometric and anthropometric investigations.<sup>2,7-9</sup> In particular, a global disharmonious appearance has been observed, with reduced lower facial dimensions, a prominent forehead and high-set orbits, maxillary and mandibular hypoplasia, narrow and short nose and ears, narrow mouth with everted lips, and an increased vermilion area of the lips.<sup>7,8,10-14</sup> HED patients showed a flat or concave facial profile with a skeletal Class III tendency (maxillary retrusion associated with mandibular protrusion) as a consequence of midface hypoplasia.<sup>5,9,13</sup>

Scant data on the palatal features in patients affected by HED can be found. Cleft palate and other palatal anomalies were recognized in some types of ectodermal dysplasia in association with ectrodactyly, midfacial hypoplasia, and ankyloblepharon. Abnormally narrow maxillary width and short palatal depth were described as characteristic of HED gene carriers by Saksena and Bixler.<sup>13</sup>

To date, no quantitative analyses of palatal shape have been performed in ED subjects. Reference data on palatal morphometry (size and shape) in subjects with HED could be useful for a better assessment of patients.

A quantitative analysis of the palate can be performed on stone casts, usually by direct techniques,<sup>15</sup> identifying landmarks on the cast and measuring them with calipers. These measuring devices are easy to use and readily available, but they are time-consuming and involve several errors and, eventually, operator fatigue. Moreover, direct anthropometry supplies linear measurements between only two landmarks at a time and neglects the third dimension, which is important for the evaluation of palatal surface. Bidimensional measurements can also be obtained with indirect techniques: casts are reproduced radiographically, or are photocopied.<sup>16</sup> The landmarks are then identified on the prints and can be digitized with a scanner and computerized measurements can be obtained, but still the third dimension is lost.

Technology offers more sophisticated systems such

as moiré stripes, stereophotogrammetry, and laser scanning to accurately describe palatal surface in three dimensions.<sup>17</sup> Unfortunately, these instruments do not provide real anatomical landmarks, and they require expensive equipment, which limits applications in large samples.

In contrast, computerized electromechanical and electromagnetic digitizers can supply the three-dimensional (3D) coordinates of anthropometric landmarks, with versatile applications, including analyses of physical forms. These methods are fast and sufficiently simple for clinical applications, and they allow creation of databases for several subsequent quantitative evaluations.<sup>18-20</sup>

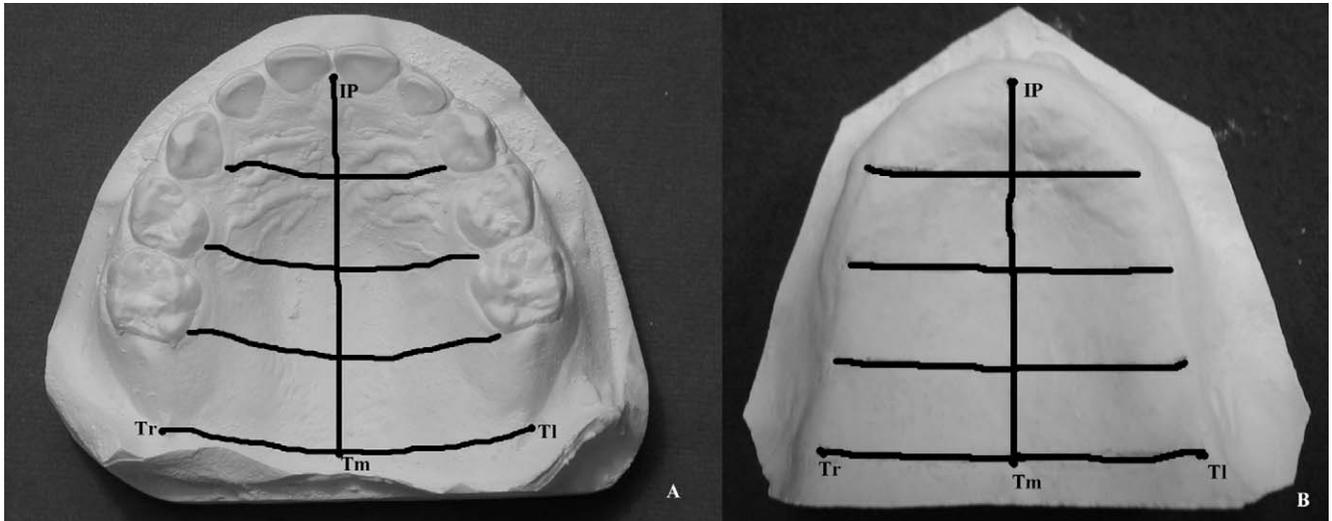
In the present study, the size and shape of the hard palate of Italian subjects with hypohidrotic ectodermal dysplasia were analyzed using a computerized 3D electromagnetic instrument that provides the spatial coordinates of actual landmarks directly digitized on dental stone casts.<sup>18,19</sup> Data were used in mathematical and geometrical models to distinguish the effect of size from that of shape on palatal features as compared with reference measures collected on healthy subjects of the same age, sex, and ethnicity.

## MATERIALS AND METHODS

Eight 6-year-old boys affected by hypohidrotic ectodermal dysplasia were analyzed in the present study. Four children (M1, M2, M3, M4) were completely edentulous and four were partially dentate (Table 1). On average, partially dentate patients had five maxillary deciduous teeth; one patient (M8) had two typically pegged first permanent molars. The crowns of all anterior teeth were conical.

The orthopantomograph revealed the germs of second maxillary permanent molars in patients M5 and M8. The mandibular arch was edentulous in six patients. Two HED boys (M6 and M8) presented a complete deciduous mandibular dentition and showed all permanent dental germs except for the first premolar teeth.

Measurements obtained from HED subjects overall were compared with data collected in our laboratory from 12 healthy Italian boys of the same age who had



**Figure 1.** Palatal landmarks marked on the stone cast of a reference boy (A) and of an edentulous HED boy (B). IP indicates incisive papilla; T<sub>r</sub> and T<sub>l</sub>, right and left limits of tuber maxillae; and T<sub>m</sub>, midpoint of the intertuber distance.

a complete deciduous dentition with erupted permanent first molars. To assess the influence of maxillary teeth on palatal size and shape, palatal measures were then calculated separately in the edentulous and partially dentate groups of HED children.

All the parents or legal guardians of the analyzed children gave their informed consent to the experiment. All procedures were noninvasive, did not provoke risks to the subjects, and were preventively approved by the local institutional review board.

Alginate maxillary impressions were taken for the HED patients during clinical activities and immediately poured in orthodontic white stone.

### Collection of Palatal Landmarks

The protocol used in previous investigations on palatal morphometry<sup>18,19</sup> was modified to avoid the complete or partial lack of dental landmarks on the alveolar edentulous crest. On each cast, the incisive papilla (IP) and the most posterior limit of the right and left maxillary tuberosity (T<sub>r</sub>, T<sub>l</sub>) were identified. The midpoint (T<sub>m</sub>) of the intertuber distance (T<sub>r</sub>-T<sub>l</sub>) was calculated and the IP-T<sub>m</sub> line traced. This line was then divided in four equidistant segments, and the relative transverse curves were traced to describe palatal morphology. On IP-T<sub>m</sub>, T<sub>r</sub>-T<sub>l</sub> and the transverse curves, 12–14 nearly equidistant points were further marked (Figure 1).

The 3D coordinates of IP, T<sub>r</sub>, and T<sub>l</sub>, and of the points on the four transverse and the anteroposterior lines were obtained by a single operator with a computerized electromagnetic digitizer (3Draw, Polhemus Inc, Colchester, Vt). The system has a resolution of 0.0005 cm/cm of range and an accuracy of 0.025 cm,

and it supplies real metric data independent of external reference systems. It consists of a transmitting electromagnetic tablet and a receiving stylus. The calibration of the system can be altered by electromagnetic interferences and metal objects; during data collection, all these devices (computer, video, and mobile phones) must be positioned a minimum of 3 m from the digitizer.

The method was found to be reliable. Repeated digitization found that for palatal landmark identification, the error percentage was about 8% of the biological variability, and for digitization, the error ranged between 1.76 and 8.26%.<sup>18</sup>

From the 3D coordinates of the landmarks, ASCII files were obtained, and computer programs devised and written by one of the authors allowed all the subsequent calculations.

To standardize and thus compare the palatal measurements for each cast, the plane described by IP, T<sub>r</sub>, and T<sub>l</sub> (X-axis: T<sub>r</sub>-T<sub>l</sub> line, right-left; Y-axis: anterior-posterior; Z-axis: caudo-cranial) was set horizontal. The following measurements were then computed for each palate:

- Sagittal plane: (1) palatal length—horizontal projection of IP-T<sub>m</sub> line (mm); (2) palatal slope—slope of the maximum palatal height vs the horizontal axis (°); (3) maximum palatal height (mm).
- Frontal plane: (1) palatal width at the maxillary tuberosity (T<sub>r</sub>-T<sub>l</sub> distance) (mm); (2) maximum palatal height (mm).

To describe palatal shape independently of palatal dimensions, all the coordinates were standardized in the sagittal plane as percentages of the horizontal pro-

**Table 2.** Palatal Dimensions in HED Patients and Reference Subjects<sup>a</sup>

Plane	Measure-ment	HED (n = 8)		Reference (n = 12)		Comparison
		Mean	SD	Mean	SD	
Sagittal	Length	34.99	3.26	39.40	1.72	*
	Slope	7.58	3.87	10.38	2.59	ns
	Height	7.84	2.34	11.66	1.27	*
Frontal	Width	40.04	3.31	40.02	3.93	ns
	Height	6.02	2.42	10.06	1.01	*

<sup>a</sup> Palatal measures are in mm, except slope (°). Comparison was made by Wilcoxon rank-sum test. HED indicates hypohidrotic ectodermal dysplasia; height, maximum palatal height in the midsagittal plane and in the intertuber line (frontal plane); length, IP-T<sub>m</sub>; width, T<sub>r</sub>-T<sub>i</sub>; and ns, not significant. \* Significant difference (*P* < .05).

jection of the IP-T<sub>m</sub> distance (y-coordinate) and in the frontal plane at the maxillary tuberosity as percentages of the intertuberosity distance T<sub>r</sub>-T<sub>i</sub> (x-coordinate).

The curvature of the palatal surface was obtained by four-degree polynomials  $y = ax + bx^2 + cx^3 + dx^4$ , both in the sagittal and frontal plane projections: in the frontal plane, the origin of the axes was set in T<sub>r</sub> (X-axis corresponding to T<sub>r</sub>-T<sub>i</sub> line; Y-axis to its vertical perpendicular); in the sagittal plane, the origin of the axes was set in IP (X-axis corresponding to the horizontal projection of IP-T<sub>m</sub>; Y-axis to its vertical perpendicular).<sup>18</sup>

**Statistical Analysis**

Descriptive statistics (mean and standard deviation) were calculated for all measurements separately for HED and control subjects. Bivariate statistic with the rectangular components of the angles was used for slope values. Comparisons between the two groups (HED and controls) were computed with nonparametric tests (Wilcoxon rank-sum test).

In HED subjects, descriptive statistics were also separately computed for partially dentate (4 boys) and completely edentulous (4 boys) palates. Data were compared by Wilcoxon rank-sum test.

A level of significance of 5% (*P* ≤ .05) was used for all analyses.

**RESULTS**

Dental formulas of the upper arch in the partially dentate HED boys are shown in Table 1. The analyzed patients presented a maximum of seven maxillary deciduous teeth, mostly canines, central incisors, and second molars.

Table 2 reports the descriptive statistics of the computed variables in the children with hypohidrotic ectodermal dysplasia and in the reference subjects. Data were compared with Wilcoxon signed rank test.

**Table 3.** Palatal Dimensions in the Two Groups of Subjects With Hypohidrotic Ectodermal Dysplasia (HED)

Plane	Measure-ment	HED Partially Dentate (n = 4)		HED Edentulous (n = 4)		Comparison
		Mean	SD	Mean	SD	
Sagittal	Length	35.78	4.08	34.40	3.00	ns
	Slope	11.67	1.66	4.92	1.51	*
	Height	10.27	0.42	6.02	0.73	*
Frontal	Width	41.87	3.21	38.68	3.04	ns
	Height	8.42	1.46	4.22	0.43	*

<sup>a</sup> Palatal measures are in mm, except slope (°). Comparison was made by Wilcoxon rank-sum test. HED indicates hypohidrotic ectodermal dysplasia; height, maximum palatal height in the midsagittal plane and in the intertuber line (frontal plane); length, IP-T<sub>m</sub>; width, T<sub>r</sub>-T<sub>i</sub>; and ns, not significant. \* Significant difference (*P* < .05).

Palatal length (IP-T<sub>m</sub>) was significantly shorter in the pooled HED patients than in the control individuals. Patients with HED showed a less steep mean palatal slope than reference subjects, but no significant difference was found because of the large intragroup variability. The maximum palatal height in both the sagittal and frontal planes was significantly lower (about 50%) in the HED subjects overall than in the control subjects. On average, HED patients and controls had similar T<sub>r</sub>-T<sub>i</sub> distance values.

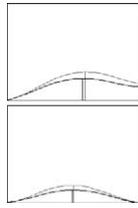
Hard tissue palatal measures in edentulous and in partially dentate HED patients are summarized in Table 3. All comparisons except palatal length and width were statistically significant (*P* ≤ .05) and partially dentate patients had all measurements larger than edentulous patients.

Differences in palatal length were minimal, about 1 mm larger in dentate HED patients.

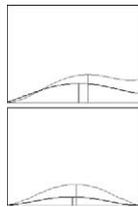
On average, partially dentate HED boys had nearly 60% steeper palates than edentulous HED boys, with a reduced intragroup variability.

In the sagittal plane, the maximum palatal height was about 42% larger in partially dentate than in edentulous patients, with a small intragroup variability. Edentulous HED boys showed a narrower palate than partially dentate HED boys, but the difference was not statistically significant.

The maximum palatal height at the maxillary tuberosity in the frontal plane was significantly modified by the presence of teeth. The edentulous children with HED had lower palates than did the partially dentate children. Overall, palatal shape was influenced by both hypohidrotic ectodermal dysplasia and edentulousness. The HED boys had a relatively lower palate than the reference boys, and in the edentulous HED boys, the hard tissue palate was relatively lower than in the partially dentate HED boys, as illustrated by the mathematical reconstruction obtained using the four-order polynomials in both projections (Figures 2 and 3).



**Figure 2.** Mean palatal shape (size-independent) in HED subjects (black lines) and reference individuals (grey lines). Frontal and lateral views. X-axis unit = % of intertuber distance; Y-axis unit = % of IP-T<sub>m</sub> distance.



**Figure 3.** Mean palatal shape (size-independent) in edentulous (black lines) and partially dentate (grey lines) HED subjects. Frontal and lateral views. X-axis unit = % of intertuber distance; Y-axis unit = % of IP-T<sub>m</sub> distance.

## DISCUSSION

Several craniofacial abnormalities have been described in subjects with hypohidrotic ectodermal dysplasia.<sup>7,8,10,12,14</sup> In particular, HED individuals present very small and retrusive malar and maxillary regions, markedly diminished lower facial depth, height, and width and a generalized reduction of the whole craniofacial complex.<sup>13</sup> Also, a nonuniform reduction of facial size, proportionally wider, produces a disharmonious appearance in HED subjects.<sup>8,14</sup> Maxillary hypoplasia is a common finding,<sup>5,13</sup> but no quantitative data on hard tissue palatal morphology are available.

In the present investigation, palatal dimensions and shape in eight 6-year-old HED boys were analyzed using an electromagnetic computerized digitizer that allows a very quick, easy, and low-cost collection of landmarks. Landmarks are digitized only once, independent of the numbers of measurements, thus reducing the potential errors of classic anthropometry data collections. Moreover, computerization permits the creation of databases and simpler data management, essential in the evaluation of large groups of subjects and for longitudinal studies. Mathematical models can also perform size-independent shape analyses.<sup>18,19</sup>

The protocol used in the current investigation was specifically devised to examine the palatal morphology independently of dental landmarks, and therefore it was suitable for both dentate and edentulous maxillary arches. Palatal measurements performed on the stone

casts of eight HED boys were compared with those of twelve reference boys using nonparametric statistics.

Overall, palatal length, slope, and maximum height in both the sagittal and frontal planes were lower in HED than in control individuals. Significant differences were found for all the cited parameters except for palatal slope because of large intragroup variability. In contrast, palatal width was similar in HED and in control boys. Reduced palatal dimensions are partially in accord with the cephalometric findings of Saksena and Bixler,<sup>13</sup> who observed that gene carriers of HED present a reduced palatal depth, but an abnormally narrower maxilla. Similarly, Bondarets and McDonald<sup>2</sup> found a smaller anteroposterior size of the maxilla (from anterior to posterior nasal spines) in lateral cephalograms of Russian ED patients.

To assess the influence of teeth on palatal morphology, HED subjects were divided in two groups: four boys were partially dentate with a mean of five teeth and four were edentulous.

All measurements were larger in partially dentate than in edentulous HED patients, with differences statistically significant except with regard to palatal length (difference of about 1 mm) and width. Therefore, palatal dimensions in HED subjects were significantly modified by the presence of teeth, as previously observed by Johnson et al,<sup>11</sup> who reported that the number of permanent maxillary teeth (erupted and pre-erupted) was significantly related to several craniofacial dimensions in patients with ectodermal dysplasia. Similar findings were reported in the 3D soft tissue analysis performed by Sforza et al.<sup>9</sup> They reported that HED subjects with a reduced number of teeth deviated more from the norm than subjects with a relatively larger number of teeth. Indeed, the dental findings reported in the analyzed HED patients were in accord with the literature.<sup>6,11</sup> In particular, the largest modification of the palate in the edentulous HED subjects occurred in the vertical dimension (palatal height and slope) in both frontal and sagittal planes as a consequence of the atrophy of the alveolar process.<sup>12</sup>

Moreover, HED boys showed palatal shape modifications such as a relative reduction of palatal height in both sagittal and frontal planes in accord with the nonuniform facial size reduction (height and depth relatively more reduced than width) assessed by Ward and Bixler<sup>14</sup> and by Sforza et al.<sup>9</sup> Also, edentulous HED boys had a relatively lower palate than partially dentate HED boys. Alteration of palatal shape could be related to the instable dental intercuspation following hypodontia/anodontia and the atrophy of the alveolar process. To obtain an appropriate mandibular posture, patients usually interpose their tongue between the dental arches during oral functions, thus altering maxillary and mandibular normal development.<sup>5</sup>

A better understanding of maxillary growth is necessary to individuate the correct timing of prosthetic rehabilitation. The treatment of pediatric patients with HED requires the clinician to have complete information about development of oral structures, behavioral management, and techniques for the fabrication of prostheses.<sup>3</sup> Today dentures are relined or rebased every 2–4 years, but prosthodontists do not possess quantitative data on the precise palatal rate of growth. Underdevelopment of the edentulous palate often leads to low retention and instability of the complete denture.<sup>3</sup> Indeed, inflammatory tissues could arise under a nonfitting prosthesis.<sup>5</sup> Alternative treatment options are overdentures supported by implants and/or by natural teeth that need ridge augmentation and vestibuloplasty for a better adaptation of the device. This therapy improves support, stability, and general psychological and social well-being. Implant positioning can maintain alveolar bone, but requires the completion of jaw development to prevent implant ankylosis. The question of the correct timing of this treatment is still controversial because of scarce knowledge of alveolar and palatal anatomy in HED patients.<sup>3</sup> Further studies in larger samples are needed to determine mean dimensions and shape of the palate in older HED patients, thus providing useful information to the clinicians during oral treatment planning.

## CONCLUSION

- Palatal size and shape were significantly modified by the presence of hypohidrotic ectodermal dysplasia, and the major alterations were found in edentulous HED subjects.

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