

Surface Morphology in Treacher Collins Syndrome: An Anthropometric Study

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Between 27 and 65 anthropometric measurements were taken for the head and face of 18 Treacher Collins syndrome patients. The most defective and frequent findings were subnormal facial depth measurements, which increased in disproportionality toward the mandible. Width of the face was more frequently and more severely subnormal than that of the mandible. The orbits were hyperteloritic with disproportionately short eye fissures and an antimongoloid inclination. The ears were microtic (more so in width than in length) in 58.8 percent of cases but low-set in only 20 percent. The nose measurements were mostly normal, which explains the "parrot-beak" character of the nose in the presence of a hypoplastic receding chin; the only nose defects were supernormal height and width in the root and subnormal bridge inclination. The most frequently seen striking disproportions were those between the width of the nasal root and the width of the soft nose, followed by the great discrepancy between the markedly narrow face and the usually normal face height.

The syndrome variously called Treacher Collins, Franceschetti-Zwahlen-Klein, or mandibulofacial dysostosis has been known for nearly 140 years (Thomson, 1846). The first recognized morphologic description was presented by Berry (1889) and the syndrome was named for E. Treacher Collins (1900) who described a major sign, the marked malar hypoplasia that produces a flattened cheek region.

Studies in this century have continued to expand our understanding of the morphologic features of the syndrome based on qualitative evaluation (Franceschetti, 1944; Franceschetti and Klein,

1949; Klein, 1953; Rogers, 1964; Klimen, 1979; Smith, 1982) and radiographic analyses of the anomaly (Stovin et al, 1960; Garner, 1967; Dahl et al, 1975; Roberts et al, 1975) (Table 1). One detailed cephalometric study of Treacher Collins syndrome has also been reported (Dagys, 1977).

These studies suggest a need for detailed anthropometric assessment of the surface disfigurements of the anomaly. In recent years the value of quantitative assessment of the direction and extent of craniofacial anomalies as an adjunct to their treatment has been recognized (Rogers, 1974; Munro, 1975; Farkas, 1981; Farkas et al, 1983). The purpose of this paper is to report the results of the preoperative anthropometric analysis of the face in 18 Treacher Collins patients seen at The Hospital for Sick Children in Toronto.

MATERIALS AND METHODS

The study sample consisted of 18 preoperative patients (9 males and 9 females) with Treacher Collins syndrome; all were white and of North American origin. Three males and three females

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TABLE 1. Qualitative Signs in Treacher Collins Syndrome Patients

Sign	%*
Antimongoloid eye fissures	89
Malar deficiencies	81
Coloboma of the lower eyelids	81
Mandibular hypoplasia	69
Lower lash deficiency	53
Ear defects	
External ear canal	40
Malformed auricle	36
Conductive deafness	28
Cleft palate	32
Tongue-shaped hair extension onto cheek	†
Relatively large nose	†
Open nasofrontal angle	†

* From Smith (1982)

† No % given

were 2 to 5 years of age, and the rest ranged from 6 to 27 years of age.

Up to 27 anthropometric measurements were taken for each patient: five on the head, 11 on the face, two at the orbits, three on the nose, four at the lips and mouth, and two on the ears. Up to 38 additional measurements were taken for the older patients: five on the head, five on the face,

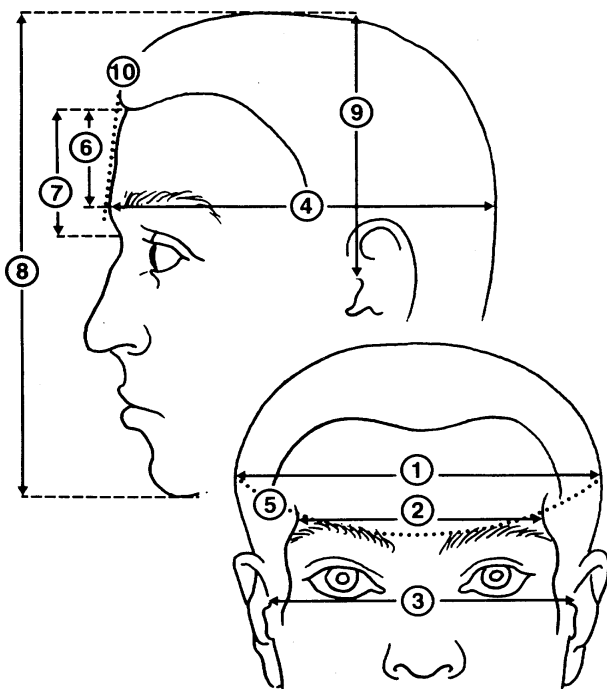


FIGURE 1. Measurements of the head: 1) head width (eu-eu), 2) forehead width (ft-ft), 3) skull base width (t-t), 4) head length (g-op), 5) head circumference, 6) height of the forehead I (tr-g)*, 7) height of the forehead II (tr-n)*, 8) combined head and face height (v-gn)*, 9) auricular height of the head (v-po, left and right)* and 10) forehead inclination (tr-g)*. (* = measurements taken only in the older group.)

six on the orbits, ten on the nose, six at the lips and mouth, and six on the ears (Figs. 1 to 6). The landmarks and technique of measurement have been described elsewhere (Farkas, 1981). Because of limitations in the control data available and problems related to the patients' ages and the degree of cooperation, not all measurements were taken for all patients.

The data for the younger patients (2 to 5 years old) were compared to norms calculated for the West German population (Hajniš, 1974). Older patients were compared to North American population norms (Farkas, 1981). Thus, more measurements were available for comparison in the older than in the younger patients.

In addition, two qualitative signs were reported: the ear was regarded as *microtic* or *rudimentary* if the measurements were subnormal; and the *vertical position* of the ear on the

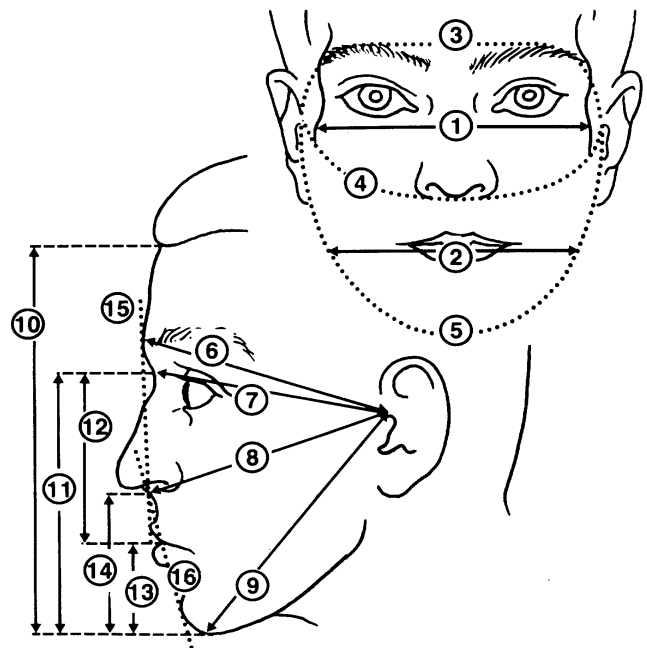


FIGURE 2. Measurements of the face: 1) face width (zy-zy), 2) mandible width (go-go), 3) supraorbital arc (t-g-t), 4) maxillary arc (t-sn-t), 5) mandibular arc (t-gn-t), 6) glabella-tragial projective distance (g-t, left and right)*, 7) upper third of face depth (n-t, left and right), 8) middle third of face depth (sn-t, left and right), 9) lower third of face depth (gn-t, left and right), 10) physiognomical height of the face (tr-gn)*, 11) morphological height of the face (n-gn), 12) upper face height (n-sto), 13) lower third of face height (sto-gn), 14) lower face height (sn-gn)*, 15) upper face inclination (g-sn)* and 16) lower face inclination (sn-pg)*. (* = measurements taken only in the older groups.)

head was established by a modification of Leiber's method (Farkas, 1978). All examinations were carried out by one of the investigators (LGF).

To demonstrate the relation between the individual measurements, 14 craniofacial indices were introduced:

1. *Cephalic index* indicates the relationship between head length (g-op) (Figure 1, #4) and breadth (eu-eu) (Figure 1, #1).
2. *Facial index* shows the proportion between morphological height of face (n-gn) (Figure 2, #11) and face width (zy-zy) (Figure 2, #1).
3. *Horizontal mandibulofacial index* presents the relation between the widths of the mandible (go-go) (Figure 2, #2) and the face (zy-zy) (Figure 2, #1).
4. *Vertical mandibulofacial index* demonstrates the proportions between the height of the lower third of the face (mandibular height, sto-gn) (Figure 2, #13) and the morphologic height of the face (total face length, n-gn) (Figure 2, #11).
5. *Jaws' arcs index* indicates the relationship between the maxillary (t-sn-t) (Figure 2, #4) and mandibular (t-gn-t) (Figure 2, #5) surface arcs.

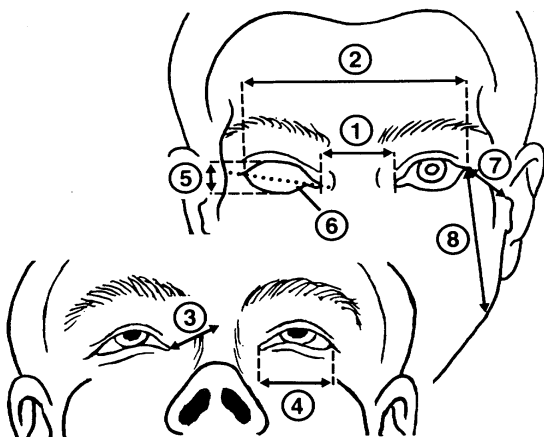


FIGURE 3. Measurements of the orbits: 1) intercanthal distance (en-en), 2) biocular distance (ex-ex), 3) endocanthion-facial midline distance (en-m, left and right)*, 4) palpebral fissure length (en-ex, left and right)*, 5) vertical palpebral fissure height (ps-pi, left and right)*, 6) palpebral fissure inclination (left and right)*, 7) orbito-tragion distance (ex-t, left and right)* and 8) orbito-gonial distance (ex-go, left and right)*. (* = measurements taken only in the older group.)

6. *Facial-craniofacial height index* relates the morphologic height of the face (n-gn) (Figure 2, #11) to the combined heights of the head and face (v-gn) (Figure 1, #8).
7. *Intercanthal index* compares the intercanthal (en-en) (Figure 3, #1) and biocular (ex-ex) (Figure 3, #2) distances.
8. *Orbital index* shows the relationship between the length of the eye fissure (en-ex) (Figure 3, #4) and the intercanthal distance (en-en) (Figure 3, #1).
9. *Nasal index* presents the relation between the width of the nose (al-al) (Figure 4, #3) and the length of the nose (n-sn) (Figure 4, #4).
10. *Horizontal nasal root-soft nose index* presents the relation between the widths of the nasal root (mf-mf) (Figure 4, #1) and soft nose (al-al) (Figure 4, #3).
11. *Nasozygomatic index* compares nose length (n-sn) (Figure 4, #4) to face width (zy-zy) (Figure 2, #1).
12. *Nasofacial index* relates nose length (n-sn) (Figure 4, #4) and morphological height of

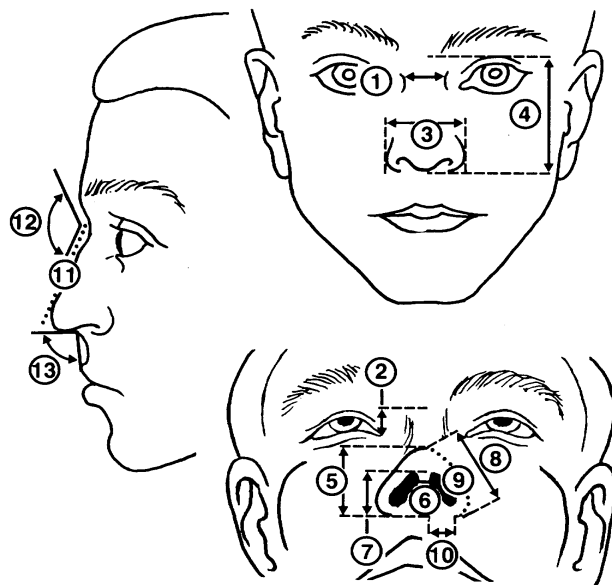


FIGURE 4. Measurements of the nose: 1) nasal root width (mf-mf)*, 2) nasal root height (en-m sag)*, 3) nose width (al-al), 4) nose length (n-sn), 5) nasal protrusion (sn-prn), 6) columella width*, 7) columella length (sn-c)*, 8) alar length, projective (ac-prn, left and right)*, 9) alar length, surface (ac-prn, left and right)*, 10) nostril floor width (sbal-sn, left and right)*, 11) nasal bridge inclination*, 12) nasofrontal angle* and 13) nasolabial angle*. (* = measurements taken only in the older group.)

face (n-gn) (Figure 2, #11).

13. *Labial index* shows the relation between the height of the medial vertical upper lip (sn-sto) (Figure 5, #3) and the width of the mouth (ch-ch) (Figure 5, #2).
14. *Auricular index* relates ear width (pra-pa) (Figure 6, #2) and length (sa-sba) (Figure 6, #1).

EVALUATION OF FINDINGS

Each measurement was classified as normal if it was within the range of the mean ± 2 standard deviations (SD) from the norms of Hajniš (1974) or Farkas (1981). Findings more than 2 SD below the mean were termed *subnormal*, while those more than 2 SD above the mean were called *supernormal*. Abnormal findings were expressed as percentages of either the minimum normal value in the case of subnormal findings (relative reduction) or the maximum normal value in the case of supernormal findings (relative increase).

RESULTS

Head Measurements

The only abnormal head measurement (Table 2) was the subnormal width of the cranial base (t-t) in two-thirds of the patients.

Head Indices

The cephalic index was supernormal in one patient, indicating a disproportionately wide head, and was above the mean but not supernormal in nine others. In the remaining eight patients the index was at or slightly below the mean.

Face Measurements

Only one of the 11 vertical facial measurements was abnormal: the height of the lower third of the face (sto-gn) was supernormal in four patients (Table 2).

Of the horizontal measurements, the width of

TABLE 2. Dimensions of the Head and Face in Treacher Collins Syndrome Patients

Measurement or Index	No. of Patients Measured	Subnormal		Mean % Difference from Minimum Normal Value	Normal		Supernormal		Mean % Difference from Maximum Normal Value
	N	%	N		%	N	%		
Head									
Cranial base width	18	12	66.7	4.1	6	33.3			
Cephalic index	18				17	94.4	1	5.6	
Face									
Lower third face height	18				14	78.8	4	22.2	15.0
Face width	18	16	88.9	9.3	2	11.1			
Mandibular width	16	9	56.3	4.8	7	43.7			
Maxillary arc	18	18	100.0	7.5					
Mandibular arc	18	16	88.9	8.5	2	11.1			
Supraorbital arc	6	5	83.3	8.8	1	16.7			
Lateral projective depth distances									
Supraorbital									
Left	2	2	100.0	4.4					
Right	2	2	100.0	7.5					
Upper third									
Left	16	11	68.8	7.1	5	31.2			
Right	16	12	75.0	6.9	4	25.0			
Middle third									
Left	16	13	81.3	7.7	3	18.7			
Right	16	14	87.5	7.3	2	12.5			
Lower third									
Left	16	13	81.3	9.2	3	18.7			
Right	16	16	100.0	7.9					
Lower face inclination	10	7	70.0		3	30.0			
Facial index	18				4	22.2	14	77.8	
Horizontal mandibulofacial index	11				5	45.5	6	54.5	
Vertical mandibulofacial index	12				8	66.7	4	33.3	
Jaws' arc index	12	1	8.3		7	58.3	4	33.3	
Facial-craniofacial height index	11	1	9.1		7	63.6	3	27.3	

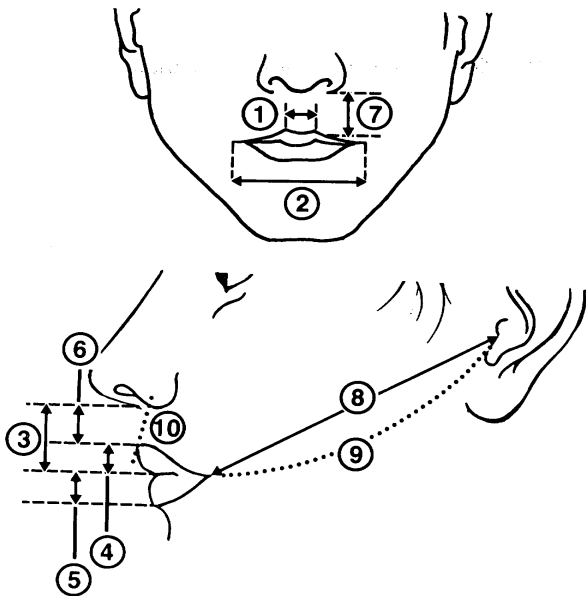


FIGURE 5. Measurements of the lips and mouth: 1) philtrum width (cph-cph)*, 2) mouth width (ch-ch), 3) medial vertical upper lip length (sn-sto), 4) upper vermillion height (ls-sto), 5) lower vermillion height (sto-li), 6) cutaneous upper lip height (sn-ls)*, 7) lateral upper lip height (sbal-ls', left and right)*, 8) labio-tragial distance, projective (ch-t, left and right)*, 9) labio-tragial distance, surface (ch-t, left and right)* and 10) upper lip inclination*. (* = measurements taken only in the older group.)

the face (zy-zy) was subnormal more often than the width of the mandible (go-go). Of the surface arcs, the maxillary arc was subnormal in all patients, and the supraorbital arc had the greatest degree of reduction (8.8%). The average reduction of the mandible was greater than that for the maxilla, but in two cases the measurement was in the normal range.

Among the lateral measurements, the projective supraorbital depth (g-t) was measured in only two adult patients and although it was subnormal in both on each side, the reduction was more pronounced on the right side. The reductions were slightly greater in the depth of the upper third of the face (n-t) and increased through the middle third (sn-t) to the lower third (gn-t).

In seven of ten cases, the inclination of the lower face profile was subnormal, which indicated a markedly receding chin.

Face Indices

The facial index (Table 2) was strikingly supernormal in 14 of 18 patients, demonstrating

a disproportionately narrow face relative to its length. In six of 11 patients the horizontal mandibulofacial index was supernormal, indicating a disproportionately wide mandible relative to face width. The vertical mandibulofacial index was supernormal in one-third of the measured subjects, showing a disproportionately elongated lower third of the face (sto-gn) in relation to face height (n-gn). The jaws' arcs index, calculated in 12 patients, was supernormal in four cases, indicating a disproportionately long maxillary arc relative to the mandibular arc. In one subject, the arcs index was subnormal.

The facial-craniofacial height index showed abnormalities in only four of the 11 patients measured. In three patients it was supernormal, indicating a long face relative to the combined head and face heights, and in one patient it was subnormal.

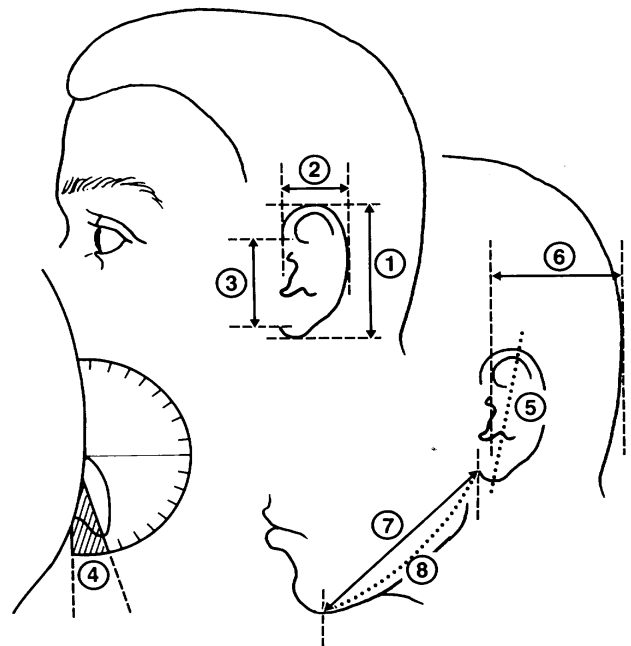


FIGURE 6. Measurements of the ears: 1) ear length (sa-sba), 2) ear width (pra-pa), 3) morphologic ear width (obs-obi, left and right)*, 4) ear protrusion (left and right)*, 5) ear inclination of the medial axis (left and right)*, 6) occipito-aural distance (op-po, left and right)*, 7) lower gnathion-aural projective distance (gn-obi, left and right)*, 8) lower gnathion-aural surface distance (gn-obi, left and right)*. (* = measurements taken only in the older group.)

Orbit Measurements

The intercanthal distance (en-en) was supernormal in only five cases, with an average of 2.8 percent over the maximum normal value (Table 3). Eye fissures were subnormal in length in more than three-quarters of those measured and had a subnormal (antimongoloid) inclination in more than two-thirds of the patients.

The lateral depths from the exocanthion to the ear (orbito-tragion distance, ex-t) and to the mandibular angle (orbito-gonial distance, ex-go) were subnormal in all patients measured. The reduction of depth was greater from the ear than from the mandibular angle.

Orbit Indices

The intercanthal index was supernormal in 13 of 18 patients, indicating hypertelorism (Table 3). The orbital index was subnormal in eight of 12 patients, showing a disproportionately short palpebral fissure in relation to the width between the eyes.

Nose Measurements

Six of ten patients had supernormal nasal root width and seven of 14 had supernormal nasal root height (Table 4). In three of 11 patients the nasal bridge inclination was subnormal and in two of ten the nasolabial angle was subnormal (acute). No supernormal angles were observed. The nasofrontal angle was supernormal in seven of ten patients, indicating a flattened ("open") angle.

Nose Indices

The nasal index ranged from subnormal to supernormal indicating great variations, from narrow and long to wide and short (Table 4). In eight of ten patients, the horizontal nasal root-soft nose index was supernormal, demonstrating a disproportionately wide root in relation to the width of the soft nose. The nasozygomatic index was supernormal in nine of 12 cases, indicating a disproportionately long nose relative to the width of the face. The nasofacial index,

TABLE 3. Dimensions of the Orbits in Treacher Collins Syndrome Patients

Measurement or Index	No. of Patients Measured	Subnormal		Mean % Difference from Minimum Normal Value	Normal		Supernormal		Mean % Difference from Maximum Normal Value
		N	%		N	%	N	%	
Intercanthal distance	18				13	72.2	5	27.8	2.8
Eye-fissure length									
Left	12	10	83.3	8.6	2	16.7			
Right	12	9	75.0	9.2	3	25.0			
Orbito-tragion distance									
Left	9	9	100.0	13.9					
Right	9	9	100.0	16.2					
Orbito-gonial distance									
Left	6	6	100.0	13.0					
Right	6	6	100.0	12.5					
Eye-fissure inclination	17	12	70.6		5	29.4			
Intercanthal index	18				5	27.8	13	72.2	
Orbital index	12	8	66.7		4	33.3			

TABLE 4. Dimensions of the Nose in Treacher Collins Syndrome Patients

Measurement or Index	No. of Patients Measured	Subnormal		Mean % Difference from Minimum Normal Value	Normal		Supernormal		Mean % Difference from Maximum Normal Value
		N	%		N	%	N	%	
Nasal root width	10				4	40.0	6	60.0	15.6
Nasal root height	14				7	50.0	7	50.0	11.8
Nasal bridge inclination	11	3	27.3		8	72.7			
Nasolabial angle	10	2	20.0		8	80.0			
Nasofrontal angle	10				3	30.0	7	70.0	
Nasal index	18	2	11.1		15	83.3	1	5.6	
Horizontal nasal root-soft nose index	10				2	20.0	8	80.0	
Nasozygomatic index	12				3	25.0	9	75.0	
Nasofacial index	12				10	83.3	2	16.7	

however, was in the normal range in ten of 12 patients, showing that the lengths of the nose and the face were proportionate.

Lip and Mouth Measurements

The mouth width (ch-ch) was slightly subnormal in five of 18 cases (Table 5). A much greater reduction was observed in the subnormal projective distances between the mouth and the ear (right and left labio-tragial distance, ch-t) on both sides in most of the patients measured. Surface measurements in the same area, taken in only six patients, were subnormal on both sides except in one patient who had a unilateral subnormal measurement. The average reduction (11%) was slightly greater than in the subnormal projective distances.

Lip and Mouth Index

The labial index was supernormal in only two of 12 patients examined, indicating a disproportionately narrow mouth in relation to the height of the upper lip (Table 5).

tionately narrow mouth in relation to the height of the upper lip (Table 5).

Ear Measurements

Nineteen auricles in the 17 patients measured were subnormal in width and one auricle was supernormal. Twenty-one auricles were subnormal in length (Table 6).

The projective and surface measurements between the lower insertion point of the ear and the chin point (gn-obi, right and left) revealed subnormal distances in all ten patients measured projectively and all five measured on the surface on the right side. Left side measurements in one subject were in the normal range.

The anteroposterior direction of the ears, determined from the occipito-aural distance (op-po), was supernormal in three of the four patients measured, indicating anterior dislocation of the ear canal (toward the face). In these patients both ears were rudimentary. In the fourth patient, the

TABLE 5. Dimensions of the Lips and Mouth in Treacher Collins Syndrome Patients

Measurement or Index	No. of Patients Measured	Subnormal		Mean % Difference from Minimum Normal Value	Normal		Supernormal		Mean % Difference from Maximum Normal Value
		N	%		N	%	N	%	
Mouth width	18	5	27.8	1.1	13	72.2			
Labio-tragial length									
Projective									
Left	9	7	77.8	11.7	2	22.2			
Right	9	8	88.9	9.0	1	11.1			
Surface									
Left	6	5	83.3	10.7	1	16.7			
Right	6	6	100.0	11.2					
Labial index	12				10	83.3	2	16.7	

TABLE 6. Dimensions of the Ears in Treacher Collins Syndrome Patients

Measurement or Index	No. of Patients Measured	Subnormal		Mean % Difference from Minimum Normal Value	Normal		Supernormal		Mean % Difference from Maximum Normal Value
		N	%		N	%	N	%	
Auricle width									
Left	17	9	52.9	26.7	8	47.7			
Right	17	10	58.8	26.0	6	35.3	1	5.9	1.1
Auricle length									
Left	17	10	58.8	14.2	7	41.2			
Right	17	11	64.7	15.2	6	35.3			
Lower gnathion-aural distance									
Projective									
Left	10	9	90.0	12.7	1	10.0			
Right	10	10	100.0	13.2					
Surface									
Left	5	4	80.0	16.3	1	20.0			
Right	5	5	100.0	13.4					
Occipito-aural distance									
Left	4				1	25.0	3	75.0	6.3
Right	4				1	25.0	3	75.0	7.0
Auricular index	17	4	23.5		11	70.6	1	5.9	

microtic right ear was shifted forward moderately.

The ears were relatively low-set on the head in three of 15 subjects, one with bilaterally dislocated rudimentary auricles and two with a low setting of one microtic ear. Five patients with bilateral rudimentary ears and three with unilateral microtia had normal ear positions in the vertical direction.

The auricular index was disproportionate in five of 17 patients evaluated. In one case, the ears were markedly wide and short (supernormal index), and in the other four patients they were disproportionately narrow and long (subnormal).

Summary of Abnormal Findings

The most frequent abnormal findings in our 18 Treacher Collins patients are summarized in Table 7.

DISCUSSION

The small size of the patient sample necessarily makes our conclusions somewhat tentative. However, the anthropometric examination confirmed the facial characteristics reported by others (Smith, 1982).

TABLE 7. Frequency of Abnormal Findings in 18 Treacher Collins Syndrome Patients

<i>Measurement or Index</i>	<i>N</i>	<i>% Abnormal</i>
Linear measurement		
Short maxillary arc	18	100.0
Short lateral depth measurements	2-16	75-100
Narrow face	18	88.9
Short mandibular arc	18	88.9
Short supraorbital arc	6	83.3
Short eye-fissure length	12	83.3
Long occipito-aural distance	4	75.0
Antimongoloid eye-fissure inclination	10	70.6
Receding lower face inclination	12	70.0
Open nasofrontal angle	10	70.0
Narrow cranial base width	18	66.7
Short ear length	17	64.7
Wide nasal root	10	60.0
Narrow ear width	17	58.8
Narrow mandible	16	56.3
High nasal root	14	50.0
Disproportion		
Wide nasal root/narrow soft nose	10	80.0
Narrow/elongated face	18	77.8
Narrow face/long nose	12	75.0
Hypertelorism orbits	18	72.2
Short eye-fissure/wide intercanthal distance	12	66.7
Narrow face/narrow mandible	11	54.5

The facial index, supernormal in 77.8 percent of the patients, showed the effect of malar deficiency in producing a disproportionately long, narrow face. The average reduction in width of the face was twice as great as in width of the mandible; this produced a marked disproportion between the widths at these two levels. Furthermore, with a few minor exceptions, all the lateral measurements were below the mean, and most were subnormal.

Disfigurement of the mandible was evident in horizontal, vertical, and surface dimensions, as well as the lower face inclination. The bigonial (lower face) breadth was subnormal in more than half of the cases (Table 2). The width of the mandibular surface arc in most cases was subnormal. In four patients the height of the lower third of the face was supernormal because of the shape deformity of the hypoplastic mandible. The greater mandibular than maxillary hypoplasia explains the supernormal jaws' arcs index.

In the orbital region, the combination of wider-than-average intercanthal distances with normal biocular distances produced a trend toward hypertelorism. The short, antimongoloid eye fissures, combined with the long intercanthal distances, aggravate the disfigurement of the entire region.

Several authors (Rogers, 1964; Gorlin et al, 1976; Klimen, 1979; Jackson et al, 1982) have indicated that Treacher Collins patients have a relatively large nose and an open nasofrontal angle. Our results indicate that the apparently large nose seems to result from abnormalities in angles, inclinations, and proportions rather than above-average dimensions of the nose. Noses with an open nasofrontal angle appear to be long. The high, wide nasal root makes the nose appear massive in the strikingly narrow face. A normal-sized nose would appear long in such a face, even if it was below an average nasofrontal angle. In addition, the vertical impression of a long nose is accentuated by the subnormal or below-average normal nasal bridge inclination. This combination of features produces a prominent nose, which has been described as "parrot-beaked" (Jackson et al, 1982).

The ears were markedly reduced in both height and width, being microtic in 71.4 percent of the cases. Although abnormal measurements were slightly more frequent in length than width, the extent of the abnormality was almost twice as

great in width measurements. The auricular index did not show any tendency for reduced width or length to predominate.

Twelve of the 17 patients had microtic ears, but only four ears were low-set when judged according to the ear canal level.

The subnormal measurements between the chin point and the lower insertion point of the ear appeared to be due to the great hypoplasia of the mandible rather than any downward dislocation of the auricle. The only true dislocation was the forward positioning of the auricles in three patients who also had microtic but not low-set ears. This forward shifting of the auricles is one cause of the subnormal projective and surface measurements between the axial facial landmarks and the ear.

The anthropometric analysis of these 18 Treacher Collins patients represents an initial attempt to quantify the characteristics of a relatively rare syndrome. Given the small sample size, very detailed analysis of age- and sex-related differences was not feasible.

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