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A Cephalometric Study of the Oral and Maxillofacial Area in Cases of CATCH 22

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CATCH 22 is the concept of a new disease proposed by Wilson and coworkers in 1993, and is an acronym for cardiac defect, abnormal face, thymic hypoplasia, cleft palate, hypocalcemia, and 22q11 deletion. The disease, CATCH 22, is characterized by 22q11.2 microdeletions, because chromosome 22q11.2 microdeletions have been confirmed as the cause of the previously known group of diseases; DiGeorge syndrome, conotruncal anomaly face syndrome, and velo-cardio-facial syndrome. We therefore conducted a study of the oral and maxillofacial area in this disease centered on an analysis of cephalometric radiograms of the head. The subjects were a total of 10 patients (mean: 15 years), and the Ricketts method was used in the analysis of the cephalometric radiograms. As a result, in the classification of facial pattern, the dolicofacial pattern was seen in 5 cases, and maxillary protrusion, mandibular protrusion or overgrowth downward, open bite, and labial inclination of the lower incisors were suggested characteristic facial findings of CATCH 22. Malformations of the first and second branchial arches during early embryonic development appeared to contribute to the morphological abnormalities of the oral and maxillofacial area in cases of CATCH 22.

Introduction

CATCH 22 is the concept of a new disease proposed by Wilson and coworkers[1] in 1993, and is an acronym for cardiac defect, abnormal face, thymic hypoplasia, cleft palate, hypocalcemia, and 22q11 deletion. The disease, CATCH 22, is characterized by 22q11.2 microdeletions, because chromosome 22q11.2 microdeletions have been confirmed as the cause of the previously known group of diseases DiGeorge syndrome[2], conotruncal anomaly face syndrome[3], and velo-cardio-facial syndrome[4].

While facial abnormalities are one of the features of CATCH 22, there has been no detailed report on a cephalometric analysis of the oral and maxillofacial area in Japan or abroad to the best of our knowledge. Cephalometric analysis has been used for orthodontics, is the base of paper surgery, and is needed for planning oral and maxillofacial surgery or plastic surgery procedures. We therefore conducted a study of the oral and maxillofacial area in this disease centered on an analysis of cephalometric radiograms of the head.

Table 1 Clinical summary of the subjects of this study

Case no. Sex, Age (y)	Cardiac defects	Complications	Other abnormalities	Mentality (IQ)
1) M, 8	TOF, PA, MAPCA, PDA (post ICR ope)	tetany (hypocalcemia), short stature	malformed auricles, nasal voice, low nasal bridge	65
2) M, 9	TOF, high AA, (post ICR ope)	tetany (hypoparathyroidism)	sinusitis, chronic otitis media, low nasal bridge	88
3) M, 9	TOF, PA, MAPCA, ARSA	hypoparathyroidism	sinusitis, otitis media with effusion	57
4) M, 13	TOF, PA, MAPCA (post ICR ope)	tetany, short stature		63
5) F, 7	IAA(B),VSD, ASD, PDA, (post ICR ope)		high arched palate, incomplete closure of the nasopharyngeal cavity	75
6) F, 10	TOF, PFO, RAA, ALSA (post ICR ope)	tetany (hypocalcemia, hypoparathyroidism)	otitis media with effusion	66
7) F, 16	TOF, PFO, RAA (post ICR ope)	tetany	facial paralysis, deafness	54
8) F, 19	RAA (post ICR ope)	hypocalcemia	malformed auricles, nasal voice, low nasal bridge, bloated eye lids	55
9) F, 29	DORV, VSD, RAA, ALSA, PH (post ICR ope)	tetany (hypocalcemia, hypoparathyroidism)	otitis media with effusion	40
10) F, 30	VSD (post ICR ope)	tetany (hypoparathyroidism) short stature, epilepsy	asymmetric crying face	45

TOF: tetralogy of Fallot, PA: pulmonary atresia, MAPCA: main artery pulmonary artery collateral artery, PDA: patentductus artery, high AA: high aortic arch, ARSA: abnormal origin of right subclavian artery, PS: pulmonic stenosis, IAA(B): interruption of aortic arch type B, VSD: ventricular septal defect, ASD: atrial septal defect, PFO: patent foramen ovale, RAA: right aortic arch, ALSA: abnormal origin of left subclavian artery, DORV: double outlet right ventricle, PH: pulmonary hypertension, ICR: intracardiac radical operation, IQ: used methods for mentality; WISC-R (case 1 ~ 4, 6, 7), TSUMORI (case 5), '87 TK Binet (case 8), WAIS-R (case 9, 10).

Subjects and Methods

The subjects were a total of 10 patients (mean: 15 years), consisting of 4 males 8 to 13 years old (mean: 9.8 years) and 6 females 7 to 30 years old (mean: 18.5 years), in whom a definitive diagnosis of CATCH 22 was made by application of the FISH (fluorescence in situ hybridization)[1] technique, in the Department of Pediatric Cardiology, The Heart Institute of Japan affiliated with Tokyo Women's Medical University.

The clinical findings of these 10 cases are shown in Table 1. All 10 patients had cardiac anomalies, including tetralogy of Fallot, and they had undergone intracardiac radical operations with the exception of 2 patients (case 3, 8). Complications included tetany (n=7), hypocalcemia (n=4), hypoparathyroidism (n=5), short stature (n=3), and similar findings. Mental development testing yielded IQ scores of 40 to 88, with a mean of 60.8, and with the exception of one patient (case 2), they all had mild to moderate mental retardation, with IQ scores below 75.

Craniofacial abnormalities consisted of sinusitis (n=2), otitis media with effusion (n=3), chronic otitis media (n=1), auricular deformities (n=2), low nasal bridge (n=3), and nasal voice (n=2) and other such problems.

The methods used in the present study centered on an analysis of cephalometric radiograms of the head and included some clinical findings

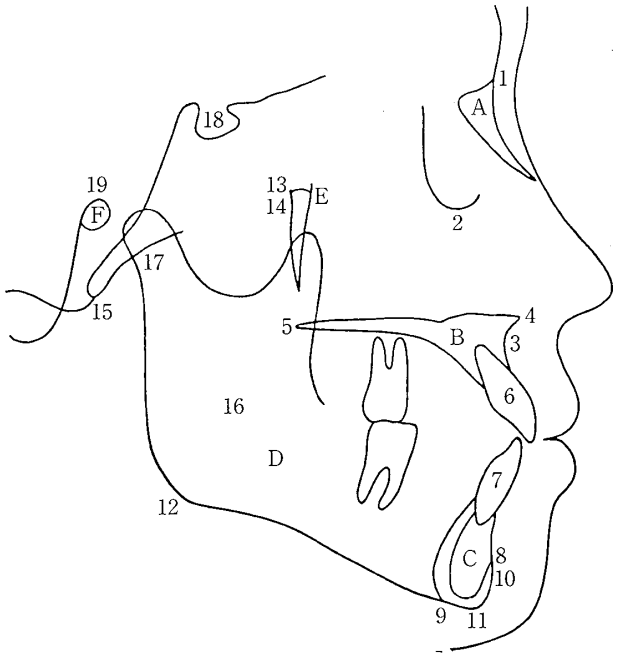

Fig. 1 Measurement points of cephalometric radiogram

A: nasal bone, B: maxilla, C: symphysis, D: mandible, E: pterygomaxillary fissure, F: external auditory meatus.

1: NA; nasion (the point at the anterior limit of the nasal and frontal sutures),
2: O; orbitale (the lowest point on the perimeter of the orbit),
3: A; point A (the most anterior point at the base of the maxillary alveolar base),
4: ANS; anterior nasal spine (tip of the anterior nasal spine of the maxilla),
5: PNS; posterior nasal spine (tip of the posterior nasal spine of the maxilla),
6: UI; upper incisor (tip of the upper central incisor),
7: LI; lower incisor (tip of the lower central incisor),
8: PM; protuberance menti (superior border of the mental protuberance),
9: ME; menton (the lowest point on the contour of the mandibular symphysis),
10: PO; pogonion (the most protruding point in the mental protuberance),
11: GN; gnathion*,
12: GO; gonion*,
13: PT; pterygoido point (inferior border of the opening of the round foramen into the posterior wall of the pterygopalatine fossa),
14: CC; Center of cranium*,
15: BA; basion (the midpoint of the anterior border of the foramen magnum),
16: XI; point XI*,
17: AR; articulare*,
18: S; sella turcica (center of the sella turcica),
19: PR; porion* (superior border of the external auditory meatus),

*: points on tracings, and point XI is the center of the ramus of the mandible on tracings and close to the mandibular foramen, anatomically. Using the plane that passes through this point makes it possible to see the morphological features of the mandible. AR is the point where the posterior border of the mandibular ramus intersects the inferior border of the temporal bone on radiogram, and GN is the point where the line that bisects the angle formed by the facial plane and the mandibular plane intersects the anterior border of the mental protuberance. GO is the point where the line that bisects the angle formed by the tangent from AR to the posterior border of the mandibular ramus and the mandibular plane intersects the mandibular border, and CC is the intersection between the BA-NA plane and the FX, with all of these points being on tracings[5)6)].

and gnathostatic model analysis.

The Ricketts method[5)6)] was used in the analysis of the cephalometric radiograms. In the Ricketts method the growth cessation age is considered to be 18 years in males and 15 years in females, and there is almost no difference between the sexes[5)6)].

The measurement points used in this study are as Fig. 1.

The following parameters were measured based on the above measurement points (Fig. 2).

The data for the above measurement parameters were assessed by measuring, analyzing, and comparing them in 10 cases of CATCH 22 patients and a control group of 27 subjects, consisting of ten 21- to 69-year-old males (mean: 37.4 years) and seventeen 18- to 69-year-old females (mean: 42.8 years). These control subjects were department staff and patients attending the outpatient clinic who had no evidence of major oral disease other than dental caries, and radiographs were taken and other examinations made only after informed consent had been obtained from the subjects themselves.

First, the patient group and Nezu and coworkers[6)7)] mean values in the Japanese population according to the age bracket were compared and tested for significant differences of one sample, and then the patient group and control group data were tested for significant differences of two samples. The Student t-test was used for statistical analysis of the cephalometric analysis data, and a value for p of less than 0.05 was deemed significant.

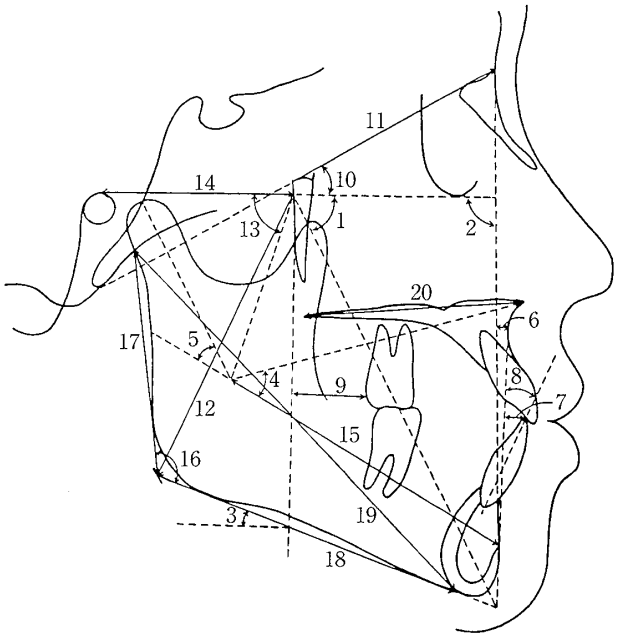

Fig. 2 Measurement parameters of cephalometric radiogram
1: FX (°); facial axis,
2: FD (°); facial depth,
3: MP (°); mandibular plane angle,
4: LFH (°); lower face height,
5: MA (°); mandibular arch,
6: PTA (mm); convexity,
7: L 1-APO (mm); mandibular incisor protrusion,
8: L 1-APO (°); mandibular incisor inclination,
9: UM-PTV (mm); upper molar position,
10: CD (°); cranial deflection,
11: CLA (mm); cranial length anterior,
12: PFH (mm); posterior facial height,
13: RP (°); ramus position,
14: PL (mm); portion location,
15: CL (mm); corpus length.

Results

The measurements and results of the analysis for 10 cases of CATCH 22 (4 males and 6 females) are shown in Table 2. The results are also traced on a Ricketts analysis chart in Fig. 3 and 4. When the mean values and standard devisions reported by Nezu and coworkers[6)7)] were regarded as normal ranges, parameters with values higher than the mean+1SD consisted of L1-APO (n=8), ∠L1-APO (n=7), MA, PTA, UM-PTV, and CL (n=6), and MP and PL (n=5). Among these values, the values exceeded the mean+2SD for L1-APO and UM-PTV (n=6), MP, MA, and PL (n=4). Parameters with values of below the mean −1SD were FX and UM-PTV (n=4), and FD and PFH (n=3). Among these values, the values were below the mean −2SD for FX and UM-PTV (n=1), FD (n=2), and PFH (n=3).

Table 2 Results of measurement

Parameters	Cases									
	1	2	3	4	5	6	7	8	9	10
FX (°)	82.9*	87.7	83.8	88.4	75.1**	92.3**	82.0*	85.9	81.2*	90.4*
FD (°)	85.8	92.0*	87.4	87.7	78.9**	92.1*	86.1	90.8	83.3*	80.5**
MP (°)	30.5	27.3	34.4*	23.4*	48.7**	26.0	39.6**	25.4	44.3**	41.0**
LFH (°)	55.8*	52.4	51.4	43.6*	57.7**	43.3*	54.0*	47.0	60.6**	49.9
MA (°)	37.3**	33.4**	17.2*	40.4**	22.6	36.4**	18.1**	31.3*	31.8*	30.3
PTA (mm)	3.7	2.2	11.3**	5.8*	11.3**	4.5	5.4*	5.6*	1.0	3.4**
L1-APO (mm)	9.3**	6.7**	5.9*	7.0**	×	6.5**	15.2**	2.8	4.6*	6.8**
∠L1-APO (°)	33.0*	34.4*	31.3*	33.4*	×	38.7**	40.6**	26.9	27.1	32.1*
UM-PTV (mm)	7.5*	8.9**	15.1**	11.6*	4.9**	23.7**	27.8**	23.1**	14.5*	26.2**
CD (°)	30.6*	29.2	32.5**	27.2	26.8	28.8	29.0	32.0*	24.7*	19.4**
CLA (mm)	58.2*	58.0	58.2*	60.0	51.0*	59.7*	62.4	58.9	59.4	64.8*
PFH (mm)	55.6	68.1**	46.4**	74.6	72.5	80.8*	76.8	76.9	74.7	74.4
PL (mm)	−33.8**	−32.8**	−37.9	−43.1*	−39.3	−37.7*	−41.6	−40.6	−33.0**	−36.6**
CL (mm)	62.9	79.6**	61.7	72.5*	53.8**	73.4**	82.2**	75.1*	76.1*	74.5

× : uneruption, *: 1SD < ≦2SD or 1SD > ≧2SD, **: >2SD or <2SD.

In addition, the following features were obtained; open bite (n=5), labial inclination of upper incisor (n=5), labial inclination of lower incisors (n=7), bimaxillary protrusion (n=4), maxillary protrusion (n=2), mandibular protrusion or overgrowth downward (n=4), hypoplasia of the hyoid bone in all cases and hypoplasia of the cervical spine (n=4). Facial pattern classification revealed 5 dolicofacial, 2 mesiofacial and 3 brachyofacial patterns. Other abnormalities consisted of enamel hypoplasia (n=1), cleft palate (postoperative) (n=4), and small mouth (n=2) (Table 3). The hyoid bone and cervical spine abnormalities were evaluated in regard to incomplete growth, developmental abnormalities, and incomplete calcification of the hyoid bone and cervical spine from the first to the fifth, comparing CATCH 22 with the cotrol group on cephalometric radiogram. Case 10 shown in Fig. 5 exhibits hypoplasia of the hyoid bone and especially the third cervical spine on the cephalometric radiogram. The cleft palate was evaluated in an interview with the patient or the patient's guardian, and the small mouth was defined as the angles of the mouth on lines perpendicular to the nasal alae or medial to them based on the standard of Kinouchi[8].

Based on Nezu and coworkers[6,7] mean values and developmental changes in the Japanese population, mean values in the Japanese according to age were set equal to 100 and the percentage of CATCH 22 patients of the same age was calculated. When the values in one CATCH 22 patient and the mean values in the Japanese were tested for significant differences of one sample, they were found for parameters L1-APO, ∠L1-APO, CLA, and PL (Table 4).

When we tested for significant differences of two samples between the CATCH 22 patients and the control group in regard to parameters FX, LFH, L1-APO, ∠L1-APO, CD, and RP, which were independent of growth or development, and between the 4 female patients over the age of 15 years, when growth or development was terminated, and the control group in regard to other measurement parameters, we found significant difference in 5 parameters; MP, PTA, ∠L1-APO, PFH, and CL (Table 5).

The representative CATCH 22 patient (case

	Clinical normal	Clinical deviation	Growth change / year
Chin in space	normal	deviation	
FX(°)	86	3	No change
FD(°)	86	3	+0.3
MP(°)	30	4	-0.2
LFH(°)	49	4	No change
MA(°)	25	4	+0.3
Maxilla			
PTA(mm)	4	2	-0.2
Teeth			
L1-APO(mm)	3	1.5	No change
∠L1-APO(°)	25	5	No change
UM-PTV(mm)	11	2	+1
Soft tissue			
LEP(mm)	2	1.5	No change
Internal structure			
CD(°)	28	2	No change
CLA(mm)	55	3	+0.8
PFH(mm)	57	4	+1.7
RP(°)	75	3	No change
PL(mm)	-39	2	-0.5
MA(°)	25	4	+0.3
CL(mm)	63	3	+1.5

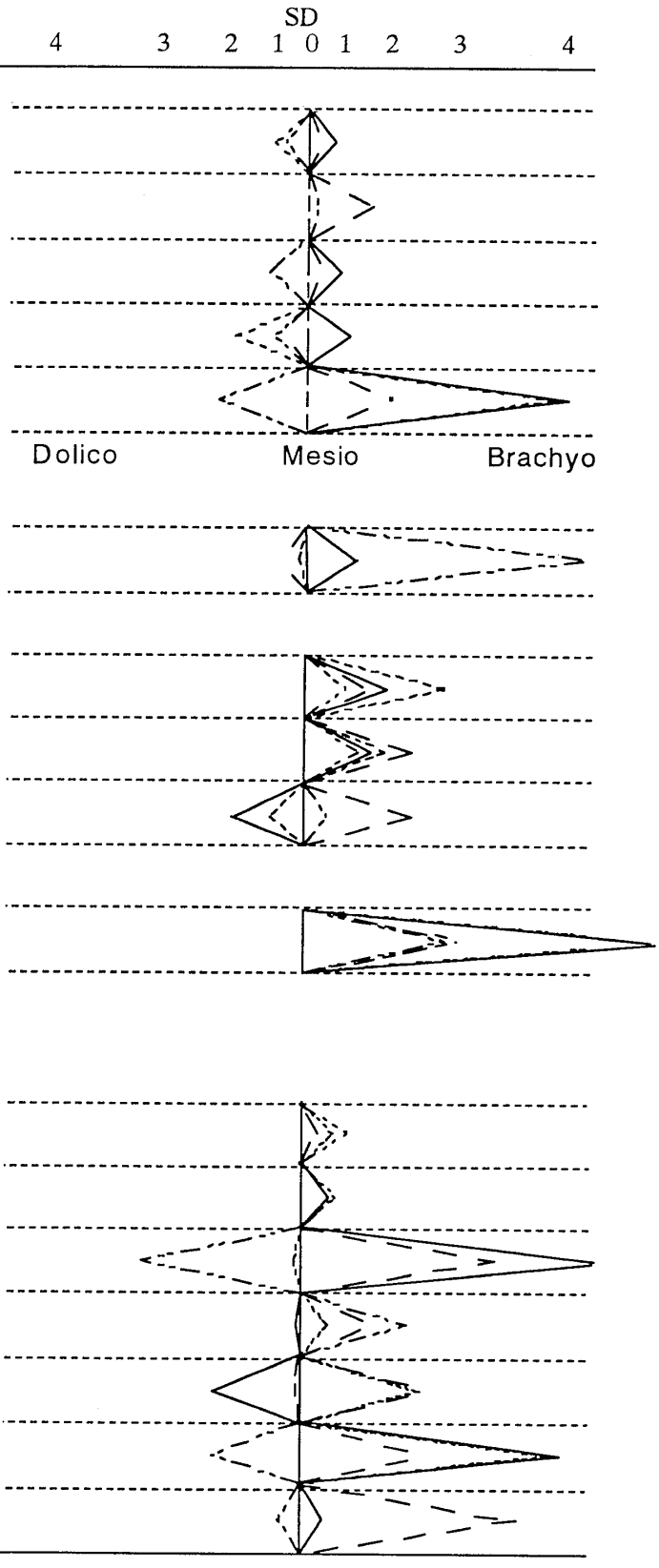

Fig. 3 Ricketts analysis (male)[6]
----- : case 1, – – – : case 2, ------- : case 3, —— : case 4.

	Clinical normal	Clinical deviation	Growth change / year
Chin in space			
FX(°)	86	3	No change
FD(°)	86	3	+0.3
MP(°)	30	4	-0.2
LFH(°)	49	4	No change
MA(°)	25	4	+0.3
Maxilla			
PTA(mm)	4	2	-0.2
Teeth			
L1-APO(mm)	3	1.5	No change
∠L1-APO(°)	25	5	No change
UM-PTV(mm)	11	2	+1
Soft tissue			
LEP(mm)	2	1.5	No change
Internal structure			
CD(°)	28	2	No change
CLA(mm)	55	3	+0.8
PFH(mm)	57	4	+1.7
RP(°)	75	3	No change
PL(mm)	-39	2	-0.5
MA(°)	25	4	+0.3
CL(mm)	63	3	+1.5

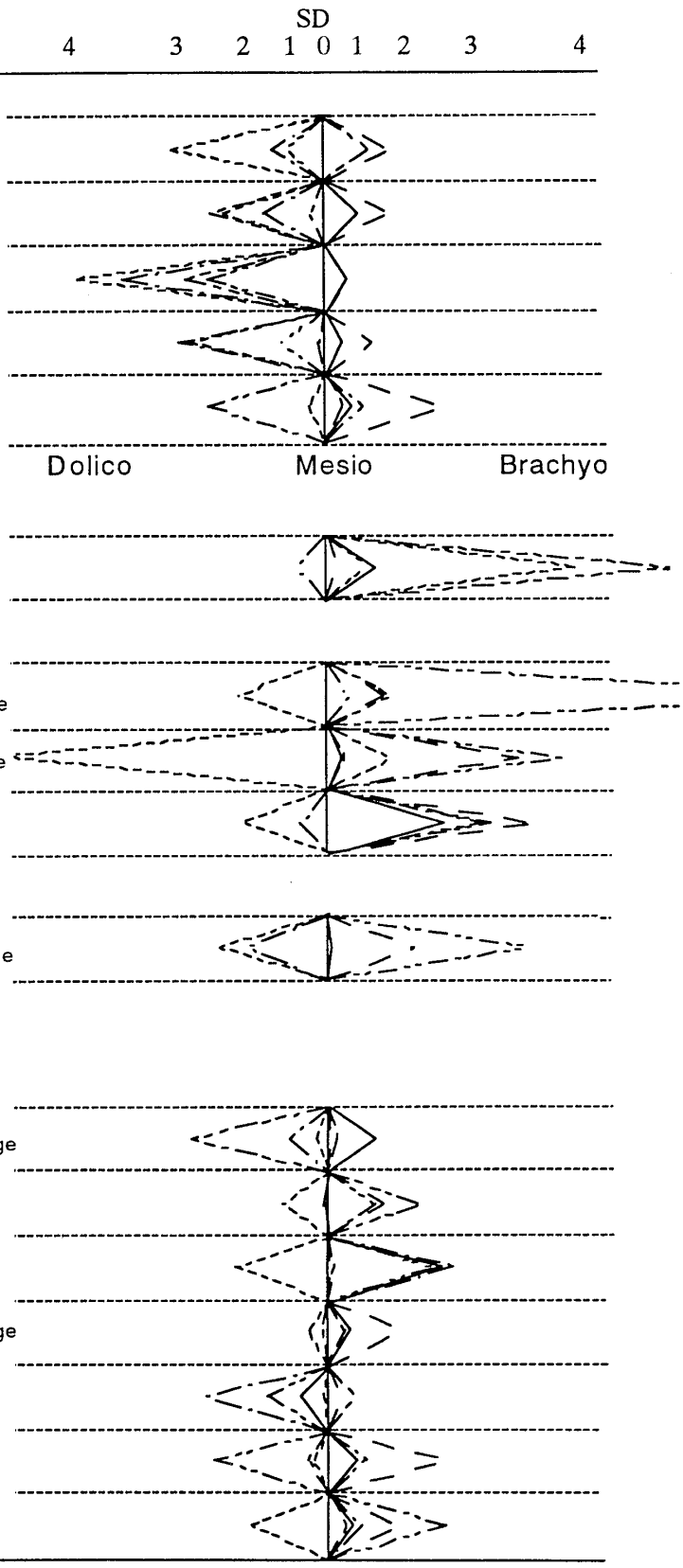

Fig. 4 Ricketts analysis (female)[6]
·····: case 5, – – –: case 6, –·–·–: case 7,
——: case 8, –··–··–: case 9, –·–·–: case 10.

Table 3 Analysis and other abnormalities

	Case no.
1) Open bite	1, 2, 5, 9, 10
2) Labial inclination of upper incisor	2, 3, 4, 6, 7
3) Labial inclination of lower incisor	1, 2, 3, 4, 6, 7, 10
4) Bimaxillary protrusion	2, 4, 6, 7
5) Maxillary protrusion	3, 10
6) Mandibular protrusion or overgrowth downward	2, 5, 9, 10
7) Facial pattern	
dolico	3, 5, 7, 9, 10
mesio	1, 8
brachyo	2, 4, 6
8) Hypoplasia of hyoid bone	all cases
9) Hypoplasia of cervical spine	2, 8, 9, 10
10) Enamel hypoplasia	5
11) Cleft palate (postoperative)	3, 8, 9, 10
12) Small mouth	1, 8

10) shown in Fig. 6 exhibits open bite, labial inclination of the lower incisors, maxillary protrusion, mandibular overgrowth downward, hypoplasia of the hyoid bone and cervical spine, and a dolicofacial pattern on the cephalometric radiogram. Facial photography is shown in Fig. 7 (case 5) and Fig. 8 (case 10). Case 5 and 10 exhibit mandibular overgrowth downward and a dolicofacial pattern on the photography.

Discussion

CATCH 22 was proposed by Wilson and coworkers[1] in the United Kingdom in 1993 as a general name for DiGeorge syndrome[2], conotruncal anomaly face syndrome[3], and velo-cardio-facial syndrome[4]. DiGeorge syndrome, reported by DiGeorge[2] in 1965, is a disease in which abnormalities of cellular immunity and hypocalcemia arise as a result of congenital aplasia of the thymus gland and parathyroid glands, and usually there are associated facial abnormalities and congenital heart defects[9]. Conotruncal anomaly face syndrome was reported by Takao[3][8][10] in 1980 and is characterized by facial abnormalities including ocular hypertelorism, mild lateral dis-

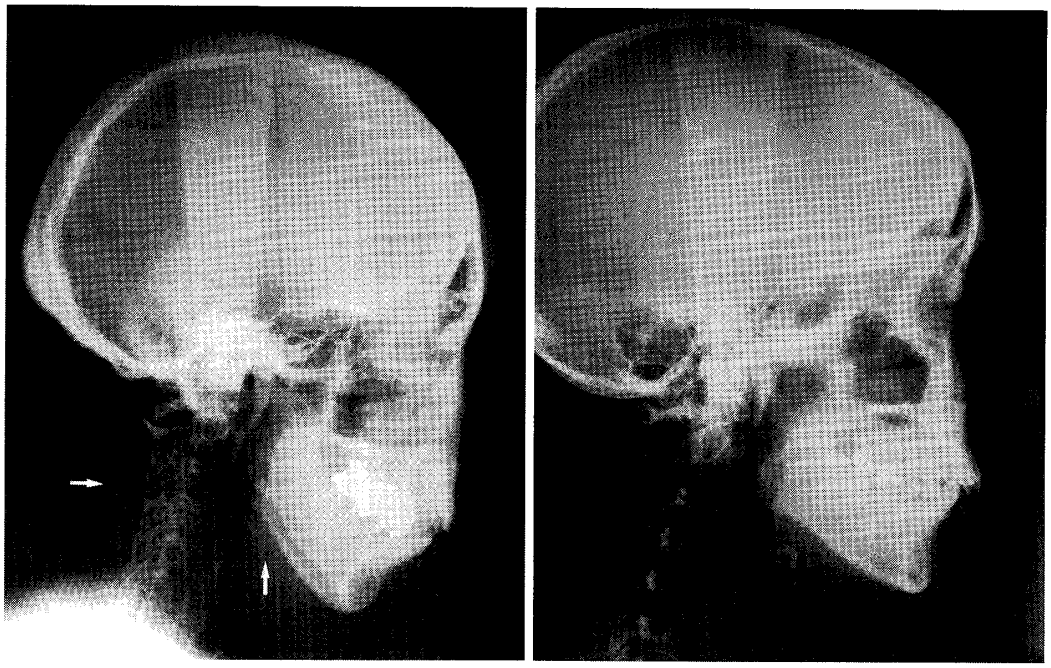

Fig. 5 Hypoplasia of hyoid bone and cervical spine on cephalometric radiogram
Left: case 10, Right: normal case.
Case 10 exhibits the incomplete growth, developmental abnormalities, and incomplete calcification of the hyoid bone and especially the third cervical spine on cephalometric radiogram.

Table 4 Comparison with 10 cases of CATCH 22 and mean values in Japanese

Parameters	Mean values in Japanese at 9 years [growth change/year] (= 100)	10 cases of CATCH 22 (CATCH 22/ mean × 100)
FX (°)	86.0±3.0	98.8 ± 5.9
FD (°)	86.0±3.0[+0.3]	99.7 ± 5.4
MP (°)	30.0±4.0[−0.2]	115.7 ± 30.2
LFH (°)	49.0±4.0	105.2 ± 11.8
MA (°)	25.0±4.0[+0.3]	116.0 ± 31.5
PTA (mm)	4.0±2.0[−0.2]	188.9 ± 130.8
L1-APO (mm)	3.0±1.5	240.0 ± 116.2*
∠ L1-APO (°)	25.0±5.0	132.2 ± 18.3*
UM-PTV (mm)	11.0±2.0[+1.0]	125.2 ± 48.7
CD (°)	28.0±2.0	100.1 ± 13.7
CLA (mm)	55.0±3.0[+0.8]	103.5 ± 4.3*
PFH (mm)	57.0±4.0[+1.7]	98.2 ± 11.0
RP (°)	75.0±3.0	102.8 ± 4.3
PL (mm)	−39.0±2.0[−0.5]	93.4 ± 8.6*
CL (mm)	63.0±3.0[+1.5]	106.3 ± 10.0

*: significant difference $p < 0.05$.

Table 5 Comparison with CATCH 22 and control

Parameters	CATCH 22	Control
FX (°)#	85.0 ± 5.0	84.6 ± 4.4
FD (°)	85.2 ± 4.4	87.4 ± 3.0
MP (°)	37.6 ± 8.4	28.7 ± 5.8*
LFH (°)#	51.6 ± 5.8	51.3 ± 5.2
MA (°)	27.9 ± 6.5	31.7 ± 5.1
PTA (mm)	6.4 ± 5.2	1.2 ± 3.8*
L1-APO (mm)#	7.2 ± 3.5	5.5 ± 3.3
∠ L1-APO (°)#	33.0 ± 4.6	27.7 ± 6.6*
UM-PTV (mm)	22.9 ± 5.9	20.0 ± 4.1
CD (°)#	28.0 ± 3.8	28.6 ± 2.4
CLA (mm)	61.4 ± 2.8	60.3 ± 3.4
PFH (mm)	63.4 ± 3.5	72.1 ± 6.5*
RP (°)#	77.1 ± 3.2	77.1 ± 3.4
PL (mm)	− 37.9 ± 3.9	− 41.7 ± 3.7
CL (mm)	77.0 ± 3.5	71.7 ± 4.1*

*: significant difference $p < 0.05$, #parameters: 10 cases of CATCH 22, other parameters: 4 cases of CATCH 22 (over the age of 15 years).

placement of the inner canthi, short palpebral fissures, single-edged and bloated eyelids, malformed auricles, low nasal bridge and small mouth: in other words, there is a combination of unique facies characterized by small eyes, a short space below the nose, a small mouth, vertically narrow face, and conotruncal anomalies, and there is often associated thymic hypoplasia, hypocalcemia, and abnormalities of cellular immunity. In many cases the syndrome is also associated with mild mental retardation and tetany occurs secondary to hypocalcemia in the neonatal period[3)10)]. In addition, incomplete closure of the nasopharyngeal cavity is observed in a high percentage of cases, and for this reason patients exhibit hinolalia apelta, and eustachian tube obstruction and otitis media tend to develop as complications[3)10)].

The difference between these two syndromes is that the principal cardiac defect in conotruncal anomaly face syndrome is tetralogy of Fallot and is complicated with a high aortic arch[11)].

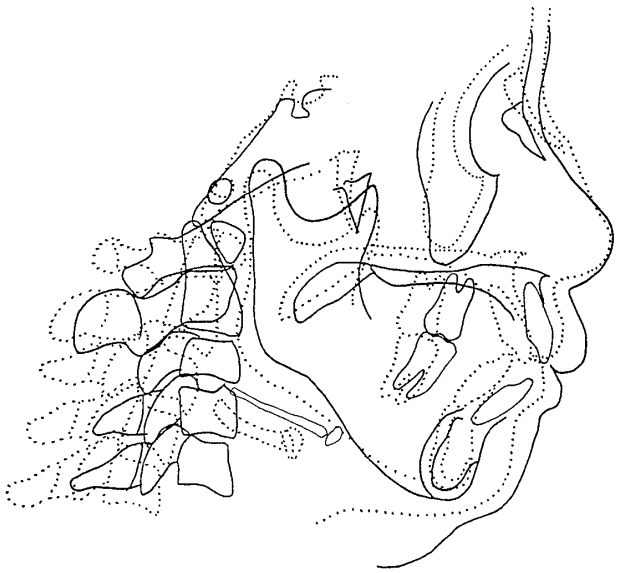

Fig. 6 The representative case of CATCH 22 (case 10)
——: case 10, ……: normal case.
Case 10 exhibits open bite, labial inclination of the lower incisor, maxillary protrusion, mandibular overgrowth downward, hypoplasia of the hyoid bone and cervical spine, and a dolicofacial pattern.

Velo-cardio-facial syndrome was reported by Shprintzen[4)] in 1978 and resembles conotruncal

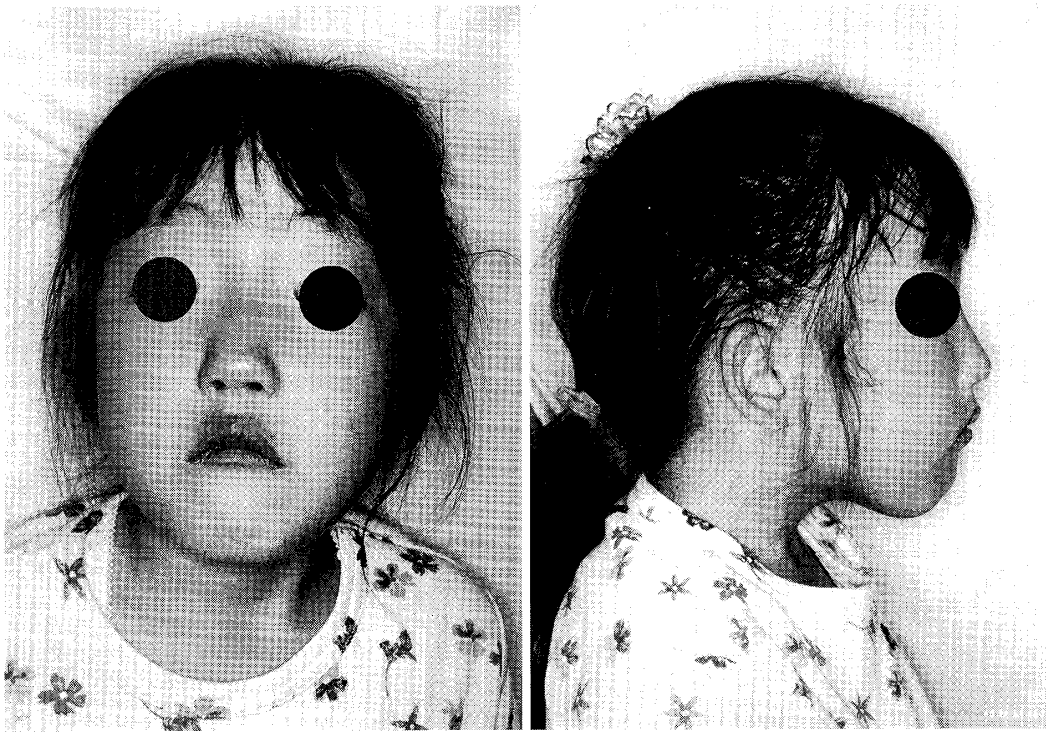

Fig. 7 The face of CATCH 22 (case 5)
Case 5 exhibits mandibular overgrowth downward and a dolicofacial pattern.

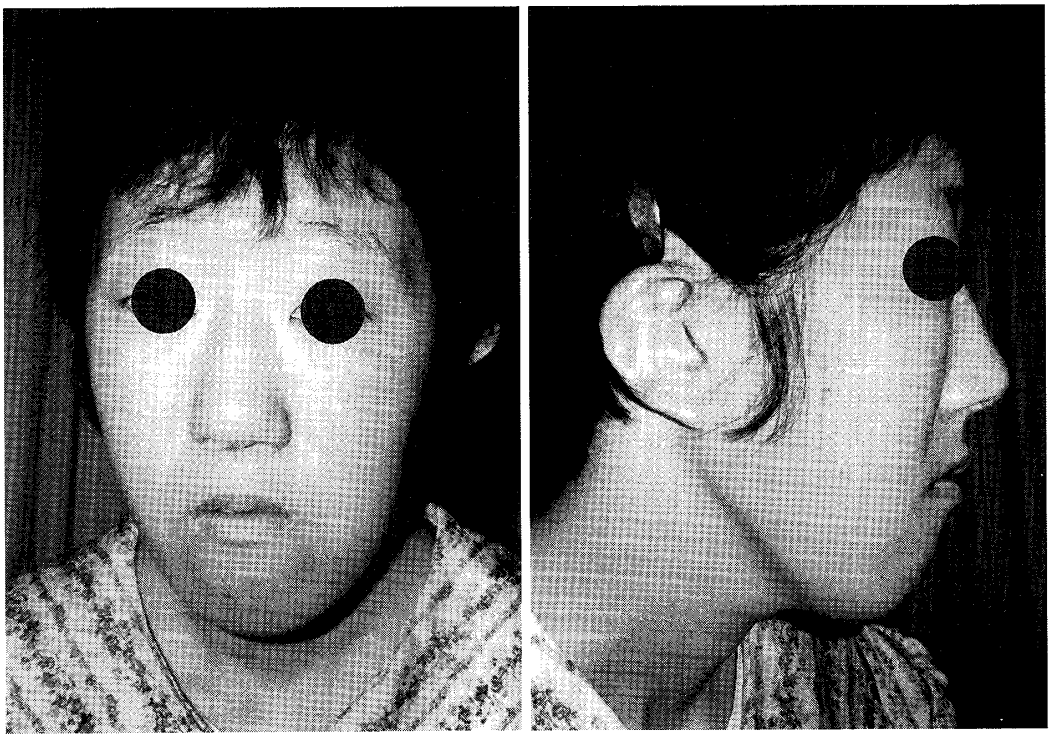

Fig. 8 The face of CATCH 22 (case 10)
Case 10 exhibits mandibular overgrowth downward, and a dolicofacial pattern.

anomaly face syndrome.

These diseases have been shown to be caused by microdeletions of the q11.2 region of chromosome 22 by application of the FISH (fluorescence in situ hybridization) molecular biology technique with DNA probes for that region[1)12)~16)].

Chromosome 22q11.2 deletions are said to occur in 1 out of 3,000～4,000 births, and they are detected in about 3% of congenital heart disease cases and about 10% of tetralogy of Fallot cases[17)18)]. That makes it the second most frequent chromosome abnormality in congenital heart disease after Down's syndrome[18)]. Since more than 90% of parents are normal, these partial deletions presumably occur when gametes undergo reduction division, and when this chromosome abnormality is present in the mother, there is said to be a 50% probability of the same abnormality occurring in her children[18)19)].

The reason that cardiac and other anomalies are observed in a high percentage of CATCH 22 cases is thought to be that malformations occur in branchial arches as a result of abnormalities in neural crest cell migration and differentiation in the early stages of development[20)]. It was assumed that malformations of the third and fourth branchial arches occurred in DiGeorge syndrome, but actually many cases are said to be associated with abnormalities of the first, second, and sixth arches[21)].

Since especially high rates of abnormalities in the growth of the maxilla and mandible, cleft palate, small mouth, hypoplasia of the hyoid bone, and so on, are observed in the orognathofacial area, they are thought to be attributable to malformations of the first and second branchial arches.

The analysis of cephalometric radiograms of the head performed in this study provided useful material for determining the morphological characteristics of the maxilla, face, and palate, and it has provided important data for a variety of clinical activities including investigating the growth of the maxilla and face, diagnosing skeletal irregularities and judging the efficacy of treatment, principally in the field of orthodontics[6)].

The results of the analysis revealed that many of the patients had large values for L1-APO, ∠L1-APO, CLA and so on, and small values for PL significantly differed from the mean values in Japanese. In addition, the facts that the value of L1-APO was large indicated protrusion of the mandibular dental arch, that the value of PL was small indicated the tendency to mandibular protrusion caused by anterior displacement of condyle, and that, the value of ∠L1-APO was large indicated labial inclination of the lower incisors. Furthermore, the fact that the value of CLA was large indicated maxillary protrusion caused by cranial length anterior was long.

In addition, the MP, PTA, ∠L1-APO, and CL values were large and PFH was small, and significantly differed from the control group. The fact that the value of MP was large indicated open bite, the value of CL was large indicated mandibular protrusion becaused by corpus length was long, and the value of PFH was small indicated a short ramus and dolicofacial pattern based on the opening mandibular angle. The fact that the value of PTA was large indicated maxillary protrusion of the skeletal pattern.

In comparison with the mean values for Japanese and for the control group, maxillary protrusion, mandibular protrusion or overgrowth downward, open bite and labial inclination of the lower incisors were considered characteristic facial findings of CATCH 22.

Conclusion

A study of the oral and maxillofacial area centered on an analysis of cephalometric radiograms was conducted in the cases of CATCH 22, and the following results were obtained:

1. Many of the patient group exhibited abnormal values for parameters L1-APO, ∠L1-APO, MA, PTA, UM-PTV, CL, MP, and PL. The values of L1-APO, ∠L1-APO, CLA, and PL significantly differed from the mean values in the Japanese.

2. There were significant differences in values of MP, PTA, ∠L1-APO, PFH, and CL between the patient group and the control group.

3. In the classification of facial pattern, there were 5 cases with the dolicofacial pattern, and maxillary protrusion, mandibular protrusion or overgrowth downward, open bite and labial inclination of the lower incisor were suggested characteristic facial findings of CATCH 22.

4. Other abnormalities consisted of hypoplasia of the hyoid bone in all cases, hypoplasia of the cervical spine in 4 cases, enamel hypoplasia in one case, cleft palate (postoperative) in 4 cases and small mouth in 2 cases.

5. Malformations of the first and second branchial arches during early embryonic development appeared to contribute to the morphological abnormalities of the oral and maxillofacial area in cases of CATCH 22.

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CATCH 22 症例における口腔顎顔面領域のセファログラム分析による検討

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CATCH 22 は，cardiac defect, abnormal face, thymic hypoplasia, cleft palate, hypocalcemia, 22q11 deletion の略で，従来の疾患である DiGeorge 症候群，円錐動脈幹異常顔貌症候群，velo-cardio-facial 症候群が染色体 22 番 q11.2 の微細欠失により生じることが確定されたため，それら 22q11.2 欠失による症候群に対して命名された疾患である．

本疾患は顔貌の異常が特徴の一つでありながら，口腔顎顔面領域の形態学的研究の報告は，国内外を渉猟してもみられない．そこでわれわれは本症例に対し，頭部 X 線規格写真分析を中心とした口腔顎顔面領域の検討を行なったので報告する．

対象は当院附属日本心臓血圧研究所小児科で CATCH 22 と確定診断された 10 例（平均 15 歳）であり，頭部 X 線規格写真分析は Ricketts 法で行なった．分析の結果は，次のとおりである．

1. L1-APO, ∠L1-APO, MA, UM-PTV, PTA, CL などに異常が多く，日本人平均値と比較し L1-APO, ∠L1-APO, CLA, PL に有意差が認められた．

2. 対照群と比較すると，MP, PTA, ∠L1-APO, PFH, CL に有意差が認められた．

3. 顔貌形態では長顔型が 5 例と半数を占め，顔貌上の特徴として，上顎前突，下顎前突，開咬，下顎前歯唇側傾斜などが示唆された．

4. 舌骨の形成不全が全例，頸椎の形成不全，口蓋裂（術後）が 4 例，小口症が 2 例，エナメル質形成不全が 1 例認められた．

またこれらの口腔顎顔面領域の形態学的異常は，発生初期の第 1，第 2 鰓弓の形成異常が関与していると考えられた．