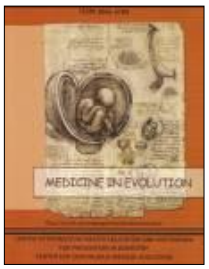
PATERN OF CEPHALOMETRIC ANALISYS AND DENTAL PROFILE IN A GIRL WITH PRADER-WILLI SINDROME



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ABSTRACT

The growth and development of craniofacial complex are multifactorial and involve genetic, environmental, endocrine, nutritional influences and result in a wide range of characteristics in healthy individuals in different populations but especially in individuals with genetic or non-genetic conditions.

The aim of the study was to evaluate the craniofacial morphology, oral, and dental findings of an 11-year and 10 month-old girl with Prader-Willi syndrome.

Key words: *Prader Willi syndrome, cephalometry*

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INTRODUCTION

Prader-Willi syndrome is associated with short stature, obesity, hypogonadism and mental retardation. The peculiar face characteristics include almond shaped eyes, thin and long upper lip and down turned corners of the mouth. This are useful for clinical diagnosis but are subjective traits.

Accurate facial analysis is essential for diagnosis of genetic and acquired anomalies, for the study of normal and abnormal growth and for morphometric investigation. Facial anthropometry plays an important role in the diagnosis of several dysmorphic syndromes and clinicians have to apply the best diagnostic techniques. The cephalometric analysis is a diagnostic tool which can provide specific and important information about the facial disharmonies also critical for the follow up of these patients.

There are only few reports about intraoral and dental features for patients with Prader-Willi syndrome; the following manifestations have been described: hypotonic orofacial muscles, high and narrow palate, hypoplastic enamel, little and sticky saliva, increased caries activity and excessive tooth.

The growth and development of craniofacial complex are multifactorial and involve genetic, environmental, endocrine, nutritional influences and resulting in a wide range of characteristics of healthy individuals in different populations but especially in individuals with genetic or non-genetic conditions [1].

The purpose of this study is to investigate the cephalo-facial relationships of the craniofacial complex in an 11-year and 10 month-old girl with Prader-Willi syndrome.

We evaluated the craniofacial growth pattern, facial profile, the relationship of apical bases with the skull, anterior and posterior facial heights, the facial height index, angular relationships formed by cranio-facial reference planes and inclinations of upper and lower permanent teeth.

CASE REPORT

The proposita is an 11-year and 10 month-old girl. Clinical examination reveals hypotonia with muscle hypotrophy, kyphotic appearance and globulous thorax.

The height was 140.8 cm, weight 66 kg and body mass index was 33.29 corresponding to class I obesity.

Diagnosis of Prader-Willi syndrome was performed by means of clinical and genetic studies. The patient had the methylation test for SNRPN gene locus on chromosome 15 and the diagnosis of Prader-Willi syndrome was confirmed. We registered aggressive attitude on frustration, limited vocabulary and articulation difficulties, the patient presents inferior retrocheilia and deep labiomenton groove.

Metric judgment criteria of craniofacial appraisal

The lines that pass through outer angle of the eyes and the mouth commissures and the facial midline revealed facial asymmetry. The angle of lower face height informs about the divergence degree and about the vertical skeletal pattern.

The profile was convex with vertical forehead and retruded chin. The nasolabial angle was found enlarged. Facial harmony was evaluated based

on the proportion between the different facial heights and the angles measurements. The horizontal lines traced through trichion and through glabella defined the upper facial part. The mid face is delimited by the horizontal lines passing through glabella and subnasale. The lower face is defined between the horizontal lines from subnasale and menton. The ideal relationship between face heights implies the equal measurements for all this parameters. By tracing the lines that separate the 3 facial heights it was established a shorter lower face height, an enlarged middle face and a brachyfacial aspect.

The lower face height might be divided into an upper third and a lower two thirds by the horizontal line passing through commissures. The balanced lower facial height requires a 1/3 to 2/3 ratio between these segments. For our patient the upper third was slightly shorter.

Facial anthropometric measurements

The patient measurements were taken with the subject sitting on the chair; head in anatomical position and they were taken to the nearest 1mm [2].

Cephalometric analysis

Cephalometric analysis is a useful diagnostic tool to determine facial type and its growth pattern, so that the clinician can determine facial disharmonies. There are several methods that can be used, but we selected two methods: craniometrical and cephalometric analysis.

For lateral cephalogram it was used a standardized radiographic technique. The cephalograms were with the head positioned parallel to the Frankfort plane, the teeth were in occlusion and the lips relaxed (figure 1).

Fig 1. Patient cephalogram.

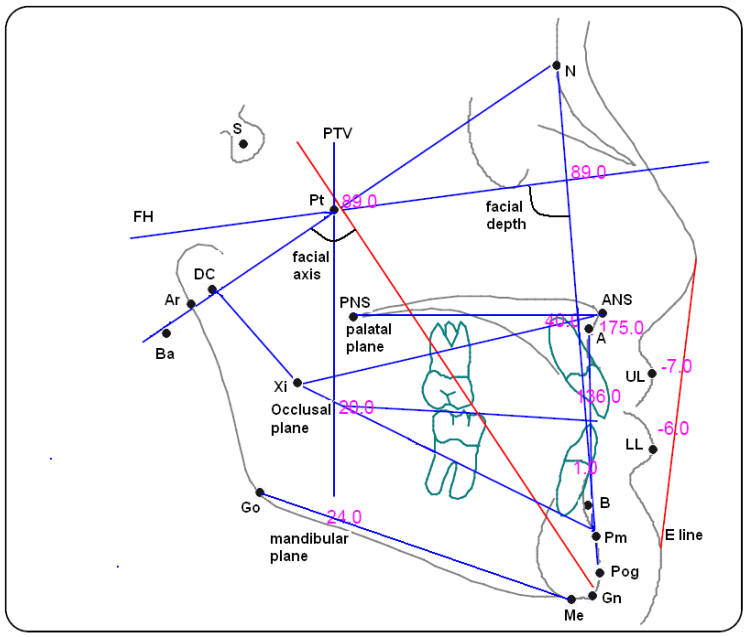

Fig 2. The landmarks, planes and parameters analyzed on the cephalometric radiographs of the patient: S (sella), N (nasion), Go (gonion), Gn (gnathion), ANS (anterior nasal spine), PNS (posterior nasal spine), A (subspinal) B (supramenton), Me (menton), Ba (basion), Pog (pogonion), Xi (mandibular center), Ul (upper lip), Ll (lower lip) and Ar (articular), Frankfort plane (FH), Palatal plane (ANS-PNS), Occlusal plane, Mandibular plane (Go-Me).

The cephalometric evaluation was performed using the automatic Cephx program. The lateral cephalometric X-ray tracing showing the landmarks planes and parameters analyzed for our patient are illustrated in figure 2. Cephalometric analysis was done according to the criteria established by Ricketts [3].

For cephalometric analysis the following measurements were evaluated:

Facial anthropometric Measurements:

The cephalic index is the breadth multiplied by 100 divided by the length of the head. An index of less than 75 means that the skull is long and narrow when seen from the top; such skulls are called dolichocephalic. An index of 75 to 80 means that the skull is nearly oval; such skulls are called mesaticephalic and are typical of Europeans. A skull having an index of over 80 is broad and short, and is called brachycephalic.

The head length was measured with a spreading caliper from glabella to inion. Head breadth was measured as the maximum transverse diameter between the two euryons using a spreading caliper. The cephalic index for this patient was established as 83 which correspond to brachycephalic type.

The prosopic/facial index was calculated using the formula: Face length x 100 / face width. Based on this index, the types of face shapes were categorized according to Banister's classification: hypereuriprosopic (very broad face) (≤ 79.9), euriprosopic (broad face) (80–84.9), mesoprosopic (round face) (85–89.9), leptoprosopic (long face) (90–94.9), hyperleptoprosopic (very long face) (≥ 95). Face length was taken as the distance between nasion and gnathion and face width as the distance between the zygoma points. Facial index was calculated as 80.7 and the patient was included as euriprosopic.

The nasal index is calculated from the following formula: Width of the nose x 100 / height of the nose. A high index indicates a broad nose and a low index a narrow nose. A nasal index below 70 is described as lepthorrhine and when above 85 it is platyrrhine. An intermediate index of 70–85 is described as messorhine. In our patient width of the nose was 4 cm, the height was 4.5 cm and the nasal index was 88.8 corresponding to the platyrrhine type. Nasolabial angle was found obtuse and it is related with retroclined incisors.

Cephalometric analysis
Skeletal Relations

Diagnosing of the facial biotype is important because each type has individual characteristics according to the dental occlusion, facial harmony, orofacial muscles, as well as shape and configuration of the craniofacial structures. In an important degree, the form of the face is determined by the position of the chin which is impacted by the cranial base, the position and the growth of condyle.

Facial biotype is the set of morphological and functional characteristics that determine the direction of growth and functional behavior of an individual face, interrelated, given by inheritance or functional disorders.

The facial biotype is determined by the VERT index (relationship established numerically, the type and amount of vertical growth of the lower third of the face, caused by posterior or anterior rotation of the jaw) and involves the five angles that relate the mandibular position: axis facial, facial depth, mandibular plane, lower facial height, mandibular arc. Within the biotype variable, the following categories are considered: mesofacial, dolichofacial and brachyfacial.

The signal is negative when the growth trend is vertical, and positive

when is horizontal. The facial pattern of each subject was classified as: dolichofacial (below -0.5), mesofacial (between -0.49 and +0.49), and brachyfacial (above +0.5). The VERT index for our patient was calculated as 0.94 and the facial biotype was brachyfacial.

The brachycephalic face, with an increased facial depth is associated with upward and forward growth of the condyle, while for dolichocephalic tendencies the condyles have an upward and backward growth.

In our patient the value for Maxilla depth was valuated by using the following parameters FH to N-A and was found to be lower (89.39) meaning a maxillary position distal to cranium base, signifying a maxillary retrusion.

Maxilla height is important in order to evaluate the proportion of middle face and the increased value found in this patient signifies a dolichofacial pattern at this level. The parameter that evaluates the pattern of facial growth includes low facial height and mandibular plane angle.

The mandibular plane angle is useful to evaluate the posterior region of the jaw and the relationship between jaw and cranium structure. A sharp angle was found in this patient sustaining an overbite or close skeletal occlusion characteristic for brachycephalic type with anterior mandibula rotation. This is in concordance with other findings of the patient: horizontal pattern of growth, brachyfacial biotype and slightly square chin. The lower face height describing the relationship between maxilla and mandibula was evaluated by the angle ANS-Xi-Pog. The value of 39.7 calculated for the patient is characteristic to a close skeletal occlusion and signifies a brachyfacial lower level and a hypodivergent profile. The hypoidvergent profile is sustained by the ratio between the posterior total face height and the anterior total face height which had a value of 64.2%, above the norm of 57% characteristic for this age.

The angle between the Palatal and Mandibulary plane was sharper than normal (20.0) indicating a short lower face height and confirming the anterior measurement discussed.

In order to evaluate the disharmony of mandibular ramus we calculate the ratio between ramus height and posterior cranial base. The cranial base should have the same measurements as ramus length, but in this patient it was found a short ramus. Even if the anterior cranial base has a normal value and the posterior cranial base was larger, the N-S-Ar angle had normal value.

Denture relationship

We have been investigated the position and the inclination of central incisors compared to A-Po line and the incisors inclination related to occlusal plane. The central upper incisors were positioned back to A-Po line (2.75 mm) meaning that they are slightly retrusive or lingualized teeth. The interincisors angle was calculated between the axes of the incisors and had a value of 136.05 indicating retrusive incisors and overbite.

Soft Tissue Analysis

Riedel [4] reported that the soft tissue profile is closely related to the skeletal and dental structures that comprise the bony profile. Burstone [5] noted that the postural position of the lower lip influences the maintenance of the original mandibular incisor position.

The angle of facial convexity, described by Down in relationship with skeletal cephalometric points was evaluated in this case by its cutaneous equivalent traced between G-Sn and Sn-Pog.

A sharp positive angle (2.61) was found for the patient sustaining a skeletal class III.

Table 1 Ricketts analysis results

Descriptor	Measurement	Mean	Standard deviation	Patient measurement
MAX DEPTH Deg	FH to N-A	95.0	5.0	89.39
MAX HEIGHT Deg	N-PTV to A	54.0	5.0	59.85
SN TO PALATAL PLANE Deg		3.0	5.0	13.93
FACIAL DEPTH Deg	FH to N-Pog	90.0	4.0	87.1
FACIAL AXIS Deg	Na-Ba to PTV-Gn	90.0	4.0	89.28
FACIAL TAPER Deg	Na-Gn-Go	67.0	4.0	69.33
MAND. PLANE Deg	FH-GoGn	28.0	3.0	24.01
CORPUS LENGTH mm	Xi to Pm	70.0	5.0	77.29
RAMUS HEIGHT mm	Ar-Go	44.0	5.0	48.1
RAMUS LENGHT mm	Go - Pg	83.7	4.6	83.11
MAND. ARC Deg	DC-Xi to Xi-Pm	27.0	5.0	22.12
A pt. CONVEXITY mm	A to N-Pog	3.0	2.0	2.61
LOW.FACE.HEIGHT Deg	ANS-Xi-Pog	47.0	5.0	39.7
MAX.1 to APo mm		6.0	3.0	2.75
MAX.6 to PTV mm		14.0		20.29
MAND. 1 to APo mm		1.0	2.0	-0.64
HINGE AXIS ANGLE Deg	DC - Go - LI	90.0	3.0	92.74
MAX.1 to MAND.1 Deg		131.0	5.0	136.05
OVERJET mm		2.5	2.0	3.34
OVERBITE mm		2.5	2.0	-1.67
UPPER LIP to E-LINE mm		-4.0	2.0	-6.92
LOWER LIP to E-LINE mm		-2.0	2.0	-5.9
ANT. CRANIAL BASE mm	N-S	71.0	10.0	75.98
POST. CRANIAL BASE mm	S-Ar	32.0	5.0	39.48
SADDLE ANGLE Deg	N-S-Ar	123.0	4.0	125.15

Table 2 Ricketts analysis results

Descriptor	Mean	Standard deviation	Patient
SNA	82.0	2.0	82.21
SNB	80.0	2.0	78.73
ANB	2.0	2.0	3.48
POGONION			2.53
ANGLE CONVEXITY (DOWNS)	180.0	5.0	175.05
WITS	0.0	1.0	-0.56

Esthetics

It was analyzed the distance between the upper and lower lip to E line defined as Ricketts esthetic line traced from Pog to Pg.

The upper lip was at -6.92 mm behind the E line suggesting a retruded

upper lip which is in line with the finding of incisors position. The distance from lower lip to esthetic plane evaluates the equilibrium of soft tissue of the lips and the profile. The lower lip was -5.9 mm behind Ricketts line sustainning the lower lip retrusion also sustained by the backward position of lower incisors.

Table 3 Skeletal Factors – Vertical and Soft Tissue Analysis (Ricketts analysis)

Descriptor	Mean	Standard deviation	Patient	Descriptor	Mean	Standard deviation	Patient
GOGN-SN	32.0	4.0	33.94	A ANGLE	90.0	4.0	90.79
OM ANGLE (SCHUDY)	20.0	5.0	15.87	NOSE	18.0	2.0	10.21
Y-AXIS (JARABAK)	67.0	3.0	66.75	U/LIP:VERT mm	3.0	3.0	2.24
Y-AXIS (DOWNS)	60.0	3.0	59.58	U/LIP:EMBR mm	5.0	2.0	2.84
JARABAK HEIGHT S-GO/N-ME	64.0	2.0	64.48	U/LIP A WIDTH mm	13.0	2.0	11.78
PL-MAND PLANE	25.0	3.0	20.0	U/LIP VERMILLION / WIDTH mm	12.0	2.0	11.98
ANT FACIAL HT (N-ANS-ME)	45	3.0	48.0	U/LIP TAPER /STRAIN mm	2.0	1.0	-0.2
				A/PT CNVXTY mm	13.0	2.0	2.61
				H ANGLE mm VARIES			5.8
				L/LIP PROTRUSION mm	1.0	1.0	1.43
				L/LIP EMBR mm	5.0	2.0	5.94
				SOFT POG mm	10.0	5.0	14.8

DISCUSSION

The study motivation is based on syndrome incidence in population, the interest for this disease on the international and now even national level, possibility of a better biological status by using therapy with growth hormone and because disharmonies in orofacial sphere can be corrected by specialists.

When performing a cephalometric analysis, it is necessary to define precisely the manner in which the many different cephalometric landmarks will be determined because it is a subjective appreciation and might influence the results. The analysis of lines and angles that define this analysis provides the clinician with the skeletal characteristics and, as a result, identification of muscular pattern. These are useful for clinician in order to evaluate the facial growth response from therapeutic procedures.

The sharp mandibular plane angle, the lower face height, the sharp angle between the palatal and mandibulary plane were found in this patient sustaining a brachycephalic biotype, close skeletal occlusion and a horizontal pattern of growth. The dental and soft tissue analysis revealed a retruded pattern and overbite.

It is important to note that overbite increases from mixed dentition to permanent dentition and decreases subsequently during maturation of untreated occlusion. While some of the measurements showed increased values (maxilla height, corpus length, interincisors angle), the majority were found lower (maxilla depth, mandibular plane angle, lower face height, distance from Max.1 to A-Po line, palatal to mandibulary plane angle, the distance from incisors to E line as well as convexity angle.

Smaller measurements of cephalometric analysis were reported by Schaedel et al. [6] in their article and this is the single reference of cephalometric investigation in Prader-Willi syndrome.

CONCLUSIONS

In this patient, the maxilla is smaller and retruded, the lower face height was shorter and we found differences at mandibula evaluation, the body was larger, the corpus length was longer, the gonial angle larger and mandibular plane angle sharper. Dental evaluation recorded lingualized upper incisors and overbite. The soft tissue analysis recorded retruded upper and lower lip. We considered important to present this case which is part of a complex project that evaluates craniofacial development of Prader-Willi patients in order to establish the growth pattern and the cephalometric parameters useful for correct dental management.

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