“Characterization of dental phenotypes and treatment modalities in Korean patients with Parry-Romberg syndrome.” - A research article analysis

Ann Treesa Paul-Augustine
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"Characterization of dental phenotypes and treatment modalities in Korean patients with Parry-Romberg syndrome." – a research article analysis.

Summary of the article

The topic discussed in this research article analysis is "Characterization of dental phenotypes and treatment modalities in Korean patients with Parry-Romberg syndrome", which was published on November 25, 2020, in the Korean Journal of Orthodontics. The link to the journal article is https://pubmed.ncbi.nlm.nih.gov/33144530/. Parry Romberg syndrome (PRS) is a rare disorder that causes a gradual and progressive atrophy and asymmetry of the facial tissues. The research detailed here was conducted by Sunjin Yim , Il-Hyung Yang and Seung-Hak Baek from the Department of Orthodontics at the Seoul National University in South Korea. They performed a retrospective longitudinal chart review study of 10 Korean PRS patients in which areas of interest were extracted from their clinical data from 1998 to 2019 to devise a novel severity index and then determined the classification of PRS patients according to this index. Based on this classification, the researchers evaluated the existence of the 5 dental phenotypes with the severity of the PRS. The authors also defined treatment modalities conducted on these patients according to the severity index. The research study concluded that the novel PRS severity index helped organize primary data for classifying the diagnosed PRS patients and subsequent charting of treatment plan taking age of the patient into consideration.

Article information

1 The title of the article is "Characterization of dental phenotypes and treatment modalities in Korean patients with Parry-Romberg syndrome."

2 The authors of the article are Sunjin Yim , Il-Hyung Yang and Seung-Hak Baek.

3 The article was published in the Korean Journal of Orthodontics. The link to the journal article is http://new.kjo.or.kr/journal/view.html?doi=10.4041/kjod.2020.50.6.407#T4

4 It was published on November 25, 2020.

5 The link to the article in PubMed is https://pubmed.ncbi.nlm.nih.gov/33144530/

The DOI of the article is https://e-kjo.org/journal/view.html?doi=10.4041/kjod.2020.50.6.407

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Study analysis

1. The study type in this research article is a retrospective longitudinal study of 10 Korean patients who are diagnosed with PRS. These patients were treated for the PRS and or followed up at the Department of Orthodontics, Seoul National University Dental Hospital (SNUDH) between 1998 to 2019.
2. The authors conducted this study to investigate the occurrence of the variable dental phenotypes and the different treatment modalities using long-term follow-up data. Parry Romberg syndrome is a rare acquired degenerative condition characterized by a slow and progressive unilateral atrophy of the facial tissues including hard tissues of the face. PRS condition is seen in females predominantly with a prevalence of 1 in 250,000 in the general population. Although the atrophy can occur in the first decade or early second decade of life, the initial age of consultation can occur in adults. A single cause is not known for the development of the PRS condition. However, the primary manifestation of PRS includes enophthalmos, dark pigmentation on the cheek or frontal area, a sharp demarcation line called coup de sabre, localized scleroderma, sunken appearance and asymmetry of the face, neurological symptom, and missing teeth. The treatment of PRS requires a multidisciplinary treatment protocol since the patient is challenged aesthetically, functionally and psychologically due to the above-mentioned characteristics. There have been numerous case reports or reviews of 1 or 2 PRS cases exhibiting variable dental phenotypes such as congenitally missing teeth, abnormal crown or root morphology, delayed eruption, and malocclusion and mostly treated by orthodontic interventions. However, these studies only included one or two cases of PRS which mentioned only the dental phenotypes present in the case reports. They also did not consider the degree of severity of the PRS patients which is highly variable. It is also known that atrophy of the involved tissues is slow and progressive and hence it is crucial to investigate the treatment modality and its timing. Hence the authors of this article aimed to investigate all the dental

phenotypes and the treatment modalities by utilizing longitudinal data and a novel PRS severity index.

3. Experimental design: This study was a retrospective one which was approved by the Institutional Review Board of the Seoul National University Dental Hospital (SNUDH). Written informed consents for participating in this study were obtained from patients. The study sample consisted of 10 unrelated Korean patients who were all diagnosed with unilateral PRS. The patient pool consisted of four male patients and six female patients. The age of the patients at the first consultation ranged from 3 years to 21 years with the mean age of 11.4 +/- 5.7 years. The patients were selected based on the following criteria:

a. Patients diagnosed with PRS.

b. Patients who were seen for treatment and follow-up at the Department of Orthodontics in SNUDH between 1998 and 2019.

c. Patients whose chart records, radiographs, and clinical photographs were available.

No control group was set up for this study. The researchers first used longitudinal data from the previous years of these patients to provide the preliminary information required to assess the facial atrophy and asymmetry. Then they devised a novel PRS severity index to determine the severity of the PRS condition based on those data. The novel PRS severity index was designed by first counting the atrophy involved areas and asymmetry involved items from the previous years of clinical data. Facial atrophy involved both soft tissue atrophy and hard tissue atrophy. Soft tissue atrophy is usually defined by the skin texture, color and volume and hard tissue atrophy is defined by the size and shape of the skeleton. However, the degree of atrophy is difficult to assess. Hence the researchers used the following facial areas to create the novel severity index. The areas selected to denote soft tissue atrophy included forehead, peri-orbital tissue, middle and lower thirds of the face. The areas selected for the hard tissue atrophy comprised of atrophy in the cranial bone, orbit-zygoma, maxilla and mandible. For facial asymmetry, the four parameters selected were difference in the facial soft tissue volume between the affected and unaffected sides, deviation of the oral commissure, occlusal plane cant, and chin point deviation. After obtaining the numbers of these areas in each patient, values of 10 and 30 was assigned to the atrophy-involved area and asymmetry-involved area, respectively. The total numbers of these areas for each patient classified the PRS patient into three types: mild (score < 140),

moderate (score between 140 and 170), and severe (score >180). Next, the researchers evaluated the occurrence of five dental phenotypes in each of these patients, namely, congenitally missing tooth, dilacerated root, tooth with short root, microdontia and delayed eruption or impacted tooth according to the novel PRS severity index. Lastly, the authors described the Tx-Mod types according to the novel PRS severity index. Six treatment modalities were defined by the researchers were the following.

a. Tx-Mod-1 - growth observation alone due to mild areas of atrophy and facial asymmetry.
b. Tx-Mod-2 - use of unilateral functional appliance.
c. Tx-Mod-3 - fixed orthodontic treatment
d. Tx-Mod-4 - growth observation due to a definite need of surgical intervention.
e. Tx-Mod-5 - use of autogenous or alloplastic graft.
f. Tx-Mod-6 - orthognathic surgery combined with fixed orthodontic treatment.

A summary of the findings and the data of these patients were presented in 5 tables which included the calculation of the PRS severity index and comparison between the results of previous studies and those of the present study on PRS.

The researchers were calibrated in that they applied the same novel PRS severity index to all the 10 patients and separate values were not reported by any of them. However, the researchers mentioned about investigating the validity and reproducibility of the novel PRS severity index if other researchers wanted to use the same in future study because these are only useful in providing preliminary data for diagnosis and treatment planning and not determining the duration and prognosis of the treatment selected.

4. Results

Distribution of the soft tissue atrophy was the middle third of the face which was seen in all the patients (100%) and lower third of the face was also involved for 80% of the patients. With regards to hard tissue atrophy, the maxilla was involved in 80% of the patients and the ramus in the mandible was affected in 60% of the patients. Facial soft tissue volume difference and chin point deviation was seen in all the patients followed by occlusal cant seen in 90% of all the patients and deviation of oral commissure seen in 80% of the patients. According to the novel severity index based on the dental phenotypes, there were

three mild PRS patients two moderate PRS patients and five severe PRS patients. The study found that in 6 patients, a total of 29 congenitally missing teeth and a total of 17 teeth with short roots was observed. I also referred to the table 3 to find out whether these two dental phenotypes were independent of each other or existed together in one individual. The table revealed that 5 PRS patients had both these dental phenotypes (50%). It was also concluded that the congenitally missing teeth and tooth with short roots were mostly seen in moderate and severe cases of the PRS patients because only 1 mild PRS (1/3), 2 moderate PRS (2/2) and 3 PRS cases (3/5) confirmed the congenitally missing teeth and teeth with short roots. To reiterate this statement further, the researchers took the average of the number of the congenitally missing teeth and tooth with short root in each of the 3 types of PRS cases and took the sum of the average numbers of missing tooth and tooth with short root in each case types. The score obtained for each case showed an increase of occurrence of these phenotypes from 1.0 in mild PRS cases to 6.0 and 6.2 in moderate and severe PRS cases respectively. This also meant that the novel PRS severity index could describe the susceptibility of the PRS patients to congenital missing tooth and tooth with short root.

The occurrence of other dental phenotypes namely, microdontia, delayed eruption and dilacerated root was seen only in 4 patients, 2 patients and 2 patients respectively and all patients had either of these 3 dental phenotypes. This also implies that the prevalence of these 3 phenotypes was relatively low. However, interestingly, the side of occurrence of all the 5 dental phenotypes showed 100% concordance on the affected side of PRS. All these findings suggest that congenitally missing teeth and teeth with short root are characteristic of the moderate and severe PRS patients.

The next group of results was regarding the distribution of the treatment modalities based on the severity of these patients. Based on the six treatment plans outlined earlier, the study presented the treatment outcomes of 4 PRS patients briefly by comparing the before and after treatment in Orthopantomographs. A combination of extraction of primary teeth, forced eruption and maintenance of space for the eruption of a permanent tooth was a part of the fixed orthodontic treatment (Tx-Mod-3) for patient #8 who had a mild PRS condition. Three patients with severe PRS condition who were willing to do the treatment had only one treatment modality in common which was the use of autogenous or alloplastic

graft (Tx-Mod-5). Out of the three severe PRS patients, two patients had multiple congenitally missing teeth in addition to the similar atrophy involved areas and asymmetry involved areas which makes them the ideal candidate for the second treatment plan namely, orthognathic surgery and fixed orthodontic treatment (Tx-Mod-6). The third severe PRS patient, in addition to the Tx-Mod-5, underwent two treatment options namely, use of unilateral functional appliance (Tx-Mod-2) and fixed orthodontic treatment (Tx-Mod- 3). It is noteworthy to mention that patient #2 who was diagnosed with severe PRS condition gave up the treatment that was recommended for him which was orthognathic surgery and fixed orthodontic treatment (Tx-Mod-6). The treatment modalities for all the other patients are summarized in the table presented in the next 3 pages.

Most of these findings imply that the treatment option for a congenitally missing tooth is based on the presence of retained deciduous tooth. If a deciduous tooth is compromised, the space for the missing tooth is maintained until a decision to replace the space with a dental implant or a bridge. Conversely, the deciduous tooth is maintained as a substitute for the congenitally missing tooth. In PRS patients with microdontia, careful communication with a prosthodontist is necessary with regards to adding resin to the mesial and distal sides of the microdont. The tooth with dilacerated root in PRS patients should be monitored carefully during orthodontic treatment through frequent radiographic evaluation as there is an increased risk of external root resorption by orthodontic forces.

It is important to note that the foundation to all these treatment modalities is orthodontic treatment. The authors of this study helped formulate a strategic plan in orthodontic treatment for the PRS patients based on the complexity of the dental phenotype situation. From the Table 1, we can see that patient # 4 (age 7 years) was recommended growth observation (Tx-Mod-1) due to mild areas of atrophy and facial asymmetry. This applies to any pre-adolescent age or an adolescent age that do not show significant atrophy and facial asymmetry. However, if PRS patients like #3, #5 and #10 aged 4 years, 11 years,

PATIENT NUMBER	AGE OF INITIAL VISIT	PRS SEVERITY	ATROPHY INVOLVED AREAS	ASYMMETRY INVOLVED AREAS	TREATMENT MODALITIES
# 1	12 Years,	Mild	Mid1/3 & lower 1/3 face + ramus	Facial STV differentiation+chin	Tx – Mod – 3 fixed orthodontic

	5M		and angle of mandible	point deviation	treatment Tx – Mod – 5 use of autogenous or alloplastic graft.
# 4	7 years, 6M	Mild	Mid1/3 + periorbital tissue	Facial STV differentiation + occlusal plane cant + chin point deviation.	Tx - Mod – 1 - growth observation alone due to mild areas of atrophy and facial asymmetry.
# 8	10 years, 2 M	Mild	Mid1/3 and maxilla	All 4 asymmetry involved areas	Tx – Mod – 3 fixed orthodontic treatment.
# 3	4 years, 4 M	Moderate	Mid1/3 & lower 1/3 face + maxilla + ramus	All 4 asymmetry involved areas	Tx – Mod – 4 growth observation due to a definite need of surgical intervention.
# 5	11 years, 2 M	Moderate	Mid1/3 & lower 1/3 face + maxilla + condyle of mandible	All 4 asymmetry involved areas	Tx – Mod – 4 growth observation due to a definite need of surgical intervention.
PATIENT NUMBER	AGE OF INITIAL VISIT	PRS SEVERITY	ATROPHY INVOLVED AREAS	ASYMMETRY INVOLVED AREAS	TREATMENT MODALITIES
# 2	21 years, 11 M	Severe	All 4 soft tissue atrophy areas involved +	All 4 asymmetry involved areas	Tx-Mod-6 - orthognathic surgery combined

			maxilla and orbitzygoma		with fixed orthodontic treatment.
# 6	3 years, 6 M	Severe (Also had 9 congenitally missing teeth)	Periorbital tissue, mid 1/3 & lower 1/3 face + maxilla and Orbitzygoma +condyle, ramus and body of mandible.	All 4 asymmetry involved areas	Tx-Mod-5 - use of autogenous or alloplastic graft. Tx-Mod-6 - orthognathic surgery combined with fixed orthodontic treatment.
# 7	10 years, 6 M	Severe	All 4 soft tissue atrophy areas involved + maxilla and orbitzygoma + ramus, body, angle of mandible.	All 4 asymmetry involved areas	Tx-Mod-2 - use of unilateral functional appliance. Tx – Mod – 3 fixed orthodontic treatment. Tx – Mod – 5 use of graft.
PATIENT NUMBER	AGE OF INITIAL VISIT	PRS SEVERITY	ATROPHY INVOLVED AREAS	ASYMMETRY INVOLVED AREAS	TREATMENT MODALITIES
# 9	14 years, 9 M	Severe (Also had 11 congenitally missing teeth)	Periorbital tissue, mid 1/3 & lower 1/3 face + maxilla and Orbitzygoma +condyle, ramus	All 4 asymmetry involved areas	Tx-Mod-5 - use of autogenous or alloplastic graft. Tx-Mod-6 - orthognathic

			and body of mandible.		surgery combined with fixed orthodontic treatment.
# 10	17 years, 3 M	Severe	Forehead, mid 1/3 & lower 1/3 face +maxilla + condyle, ramus, angle and body of mandible.	All 4 asymmetry involved areas	Tx – Mod – 4 growth observation due to a definite need of surgical intervention.

17 years respectively, seek consultation before completion of growth and show significant atrophy and asymmetry, the best treatment option is growth observation for surgery (Tx-Mod-4). During the adolescent growth period, treatment options may include active use of unilateral functional appliance (Tx-Mod-2) and fixed orthodontic treatment (Tx-Mod-3) in mild to moderate PRS patients. Based on the severity and degree of atrophy and asymmetry, the treatment modalities can also extend to orthognathic surgery (Tx-Mod-6) and grafting (Tx-Mod-5) that will help in the restoration of the soft tissue deformities and reconstruction of the skeletal framework.

5. Conclusions

The authors concluded that all dental phenotypes presented in PRS patients occurred on the same side as the side affected by PRS. They also deduced that congenitally missing tooth and short rooted tooth are associated with moderate and severe PRS patients than the other dental phenotypes. The treatment modality selected for each of these patients was either a single option or a combination of many and this selection was based on the age of the patient and the classification of the PRS patient.

The researchers think that the present study only provides basic information on the dental phenotypes and Tx-Mod types in PRS patients. Hence it would be better to conduct a prospective study with a larger sample size from nation-wide multi-centers in the future. The authors also emphasize the need for investigating the validity and reproducibility of the PRS index in future study.

6. Impression

I feel this study is important with regards to our profession as dental hygienists. Knowing the primary manifestations of this rare syndrome at an early stage will alert us to create awareness in the patient of this condition and the need for consultation as soon as possible to determine its severity and the prognosis. Also, this condition is a slow, progressive atrophy of the face and may not be evident physically. Therefore, it is important to evaluate the radiographs for the presence of one or a combination of any of the five main dental phenotypes. However, convincing these patients will be challenging when the patient is aware of the syndrome and refuses the treatment either due to cost of treatment or fear of the surgical approach. This study presented an adult patient aged 21 years who gave up the treatment that was proposed to him. This patient was diagnosed with a severe PRS condition and yet refused to come for the treatment. This raises a question as to how to overcome the patient's refusal to a treatment especially when a surgical approach is recommended. This leads me to relate to one of my patients who was diagnosed PRS in her early childhood. Everything about her was normal except for the coup de sabre on one side of the face. On asking her about the latest update on her syndrome, she casually replied that she had a surgical treatment that involved the placement of an implant in the skull. I was shocked to hear it and wanted to hear about how she managed that treatment. However, she was reluctant to discuss the information at that time probably because she was pregnant on her first visit with me. Now that I met a pregnant PRS patient, I am curious to know how it will affect pregnancy.

7. References

Yim, Sunjin et al. "Characterization of dental phenotypes and treatment modalities in Korean patients with Parry-Romberg syndrome." *Korean journal of orthodontics* vol. 50,6 (2020): 407-417. doi:10.4041/kjod.2020.50.6.407