


Original Research Article

A retrospective study on tongue malformations and a suggested classification

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Abstract – Objective: The objective of this study was to present the tongue abnormalities associated with different genetic disorders as well as propose a new classification for these anomalies. **Materials and methods:** In this retrospective study, the oro-dental reports from the outpatient clinic of the Oro-Dental Genetics department from the period of 1-1-2016 till 31-12-2016 were reviewed for the tongue abnormalities present. **Results:** The study included 304 patients; 121 patients had tongue abnormalities (39.8%). Tongue size abnormalities were present in 34 patients (28%). Position abnormalities accounted for 14 cases (11.5%). Tongue coating abnormalities were present in 19 patients (15.7%). Errors of fusion particularly bifid tip of the tongue was present in 28 patients (23.1%). There was a tongue hemangioma that presented in a patient with kindler syndrome. There were other abnormalities that were suggestive of diagnoses as the hamartomatous masses associated with oro-facio-digital syndrome and others. **Conclusion:** This study described different tongue anomalies seen in a group of patients diagnosed with genetic syndromes. Also, a newer classification system for tongue anomalies was proposed, which in our point of view will aid clinicians in diagnosis of genetic disorders associated with such anomalies.

Introduction

The tongue is a particularly remarkable organ in the oral cavity, it acts as a mirror reflecting the oral and systemic health [1]. The tongue was even considered in the ancient medicine by the Greek philosophers, Hippocrates and Galen as the barometer of health [2].

A number of congenital anomalies affect the tongue which involve discrepancies in normal size, shape, position or texture of the tongue. They are structural defects that occur during embryogenesis and are identified at birth [3].

Tongue anomalies can occur either in isolation or as a clinical feature in a genetic syndrome [4]. Thus, these anomalies need to be addressed carefully, because along with other extraoral manifestations they can help reach an early diagnosis of rare genetic disorders [5].

Macroglossia, the long-term painless enlargement of the tongue is usually a sign of an underlying condition, either congenital or acquired. Isolated macroglossia is very rare (6).

Macroglossia can be either true or relative macroglossia, according to the size of the tongue in relation to the other structures in the oral cavity [6].

In true macroglossia, there is an apparent enlargement of the tongue due to an underlying condition with relevant histopathological findings. One prominent example is Beckwith Wiedemann syndrome (BWS), an overgrowth syndrome with macroglossia as one of its main features. The underlying cause of macroglossia in BWS remains unknown, however, Oyama Yuzu *et al.*, 2020 attributed the BWS-associated macroglossia to skeletal muscle hyperplasia, where histologic examination of autopsied tongue specimen revealed an increase in the number of skeletal muscle fibers [7].

While, in relative macroglossia, the tongue appears larger when compared to other structures in the oral cavity. For example, Down syndrome, where tongue appears enlarged due to hypotonia and Pierre Robin syndrome due to micrognathia [6].

Other syndromes associated with macroglossia are Mucopolysaccharidoses (Hunter syndrome and Hurler syndrome), Robinow syndrome, Maroteux – Lamy syndrome and Crouzon syndrome [6,8].

On the other hand, microglossia is an exceedingly rare developmental condition that refers to an abnormally underdeveloped tongue. It is a main feature in oro-mandibular limb hypogenesis syndrome [9].

Another example of tongue malformation is the bifid tip of the tongue. Other synonyms are also used to describe this malformation as accessory tongue, double tongue, and supernumerary tongue [10].

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Bifid tip of the tongue is rarely an isolated finding, it is usually associated with syndromes, the most commonly reported are Oro-Facio-Digital type I, II, IV, and VI, Klippel-Feil anomaly, Larsen anomaly, Goldenhar syndrome, and Ellis-van Creveld syndrome [11–14].

Ankyloglossia or tongue tie, is another congenital anomaly characterized by unusually short lingual frenum that restricts tongue mobility. The most common type of ankyloglossia is ankyloglossia inferior, it can be an isolated finding or part of syndrome as Ehlers Danlos syndrome, BWS, orofacial digital syndrome, or x-linked cleft palate [15].

Ankyloglossia superior is another type of ankyloglossia, in which the tongue tip is congenitally adherent to the hard palate, it is quite rare with only few cases reported in the literature. When ankyloglossia superior is found with other congenital anomalies, such as limb deformities and cleft palate, it is termed as ankyloglossia superior syndrome [16].

Tongue muscle weakness as well as morphological changes due to muscle hypertrophy or atrophy are often seen in patients with neuromuscular disorders. For example, in Duchenne muscular dystrophy (DMD), enlargement of the tongue is frequently observed while in amyotrophic lateral sclerosis (ALS), atrophy of the tongue is commonly seen [17].

The aim of this study was to shed light on the tongue anomalies and their importance in the early diagnosis of rare genetic diseases, subsequently there was this compelling need to update the existing classification system for tongue anomalies and propose a newer and a more detailed classification with a broader perspective for better understanding of tongue anomalies and help reach a proper diagnosis.

Materials and methods

In this retrospective study, the oro-dental reports from the outpatient clinic of the oro-dental genetics department for a period of one year were reviewed for the tongue abnormalities present and were correlated with the diagnoses. All patients signed an informed consent form to take part in this study.

The abnormalities were grouped under 10 categories: abnormalities of number, size (Fig. 1), position, errors of fusion, tongue coating abnormalities, abnormal range of movement, abnormal muscle substance, neurological manifestations, vascular lesions and masses (Tab. I).

Results

The study included 304 patients; 121 patients had tongue abnormalities (39.8%). Several abnormalities could be found in one patient. Tongue size abnormalities were present in 34 patients (28%). Particularly macroglossia was present in 33 patients and one patient had tongue asymmetry. Position abnormalities accounted for 14 cases (11.5%) where; ankyloglossia was present in 11 patients, retroglossia in 2 and glossoptosis in 1 patient (Tab. II).



Fig. 1. Long tongue.

Tongue coating abnormalities were present in 19 patients (15.7%); 8 with increased tongue coating and 11 with geographic tongue. Abnormal fusion was present in 28 patients (23.1%) with bifid tip of the tongue.

There was a tongue hemangioma that presented in a patient with kindler syndrome (Fig. 2a). There were other abnormalities that were suggestive of diagnoses. Among these abnormalities were the presence of fibrotic masses on the tongue which was present in two patients and directed the diagnosis to oro-facio-digital syndrome (Fig. 2b).

Another abnormality was limitation of tongue movement which was present in patients of epidermolysis bullosa and lipid proteinosis (Figs. 2c and 2d). Excessive protrusion was viewed in cutis laxa syndrome.

Abnormal tongue muscle was a total of 29 patients (23.9%). Tongue fissuring represented 26 patients (21.4%) with 8 patients under 20 years of age with multiple fissuring and 18 with a deep median grooved tongue.

The deficient substance of the tongue muscles which could be in the form of lingue cochlearis also known as spoon tongue or it could just be in the form of a “thin tongue” which is seen in arthrogyriposis multiplex syndromes (Fig. 3). There was a single case with muscular discrepancy in the form of 2 lateral grooves on the tongue borders.

Discussion

Tongue abnormalities especially those that are congenital can aid in the diagnosis of genetic disorders but not without the correlation of the other presenting features. Classification and grouping of the abnormalities can aid in the process of diagnosis and cataloging of the cases. In this study a suggested classification was used, from our experience it can facilitate research and can allow for the addition of syndromes for a full comprehensive reference [18]. The classification is an extension of the work done by Emmanouil-Nikoloussi and Kerameos-Foroglou, 1992, but in a more comprehensive manner that include acquired and congenital conditions [2].

Table I. New suggested classification for tongue malformations.

Category	Abnormality
Tongue number	Aglossia
	Accessory tongue
Tongue size	Macroglossia
	Microglossia
	Long tongue
	Asymmetry of the tongue
Position	Retroglossia
	Ankyloglossia
	Glossoptosis
Error of fusion	Bifid tip of the tongue
	Pentafid
Coating abnormalities	Glazed tongue
	Median rhomboid glossitis
	Increased tongue coating
	Hairy tongue
	Strawberry tongue
Abnormal muscle substance	Spoon tongue
	Tongue fissuring
	Median grooved tongue
	Thin tongue
	Wooden-like
	Mutilated
Neurological abnormalities	Tremors
	Fasciculations
	Deviation
	Dyskinesia
Range of motion	Limited
	Excessive
Vascular lesions	Hemangioma
	Vascular malformations
Masses	Lingual thyroid nodule
	Cysts
	Tumors

It is worth noting that the classification herein unlike the ICD-11 uses the abnormalities as mean of categorization not the anatomical regions. The regions can be added if needed in categories as the masses or vascular lesions [19].

Macroglossia was the most common tongue abnormality present in our sample. The presence of macroglossia doesn't always mean Down syndrome, Beckwith Weidmann or hypothyroidism, it could be isolated [20].

The most common group of findings was the errors of fusion group and bifid tip of the tongue came in second place to macroglossia. The bifid tip of the tongue is a feature of certain disorders as Ellis-van Crevald syndrome and Kabuki syndrome [21,22]. Bifid tip of the tongue can be due to high insertion of a short lingual frenum that might cause partial ankyloglossia thus a proper examination of the lingual frenum is mandatory.

Retroglossia is a term that describes the posterior position of the tongue in the floor of the mouth. It has been observed in a patient with campomelic syndrome in association with

Table II. Results of tongue abnormalities.

Abnormality group	Tongue abnormality	Number of patients
Size	Macroglossia	33
	Asymmetry	1
Position	Ankyloglossia	11
	Retroglossia	2
	Glossoptosis	1
Errors of fusion	Bifid tip of the tongue	28
Tongue coating	Increased	8
	Geographic tongue	11
Muscle substance	A deep median groove	18
	Multiple fissuring	8
	Spoon	1
Range of movement	Lateral lines on the border	1
	Thin	1
	Limitation of the tongue movement	2
	Hyperextensibility of the tongue	1
Neurological	Fasciculations & tremors	7
Masses	Fibrotic masses	2
Vascular	Hemangioma	1

retrognathia [23]. Both patients with retroglossia in this study had retrognathia as well. One of the patients had proportionate short stature which could mean that the tongue presented a degree of microglossia. The other patient had retroglossia associated with retrognathia and ankylosed temporomandibular joint.

Excessive tongue coating could be attributed to the patients' inability to keep an oral hygiene regimen which is the first determinant of tongue coating, secondly comes poor food intake [24]. In the study herein, it was also seen in association with the fibromas in the patient with OFD.

As for the range of motion it is either limited or excessive. It goes without saying that the limitation in this classification is caused by anything other than ankyloglossia. In lipoid proteinosis the tongue is affected in both the range of motion and the substance of the muscle where the tongue becomes wooden like as the cases progress, another example of affection over several categories [25]. Epidermolysis bullosa patients often find it difficult to fully extend the tongue due to healing of the bullae with fibrosis [26].

As for the affection of the muscle substance, the term thin tongue to our knowledge it has never been used before in describing tongue abnormalities, but it is a frequent observation in the patients with arthrogryposis multiplex if spoon tongue is not fully present. It is a type of muscle hypoplasia that they exhibit, probably a mild degree of spoon tongue [27].

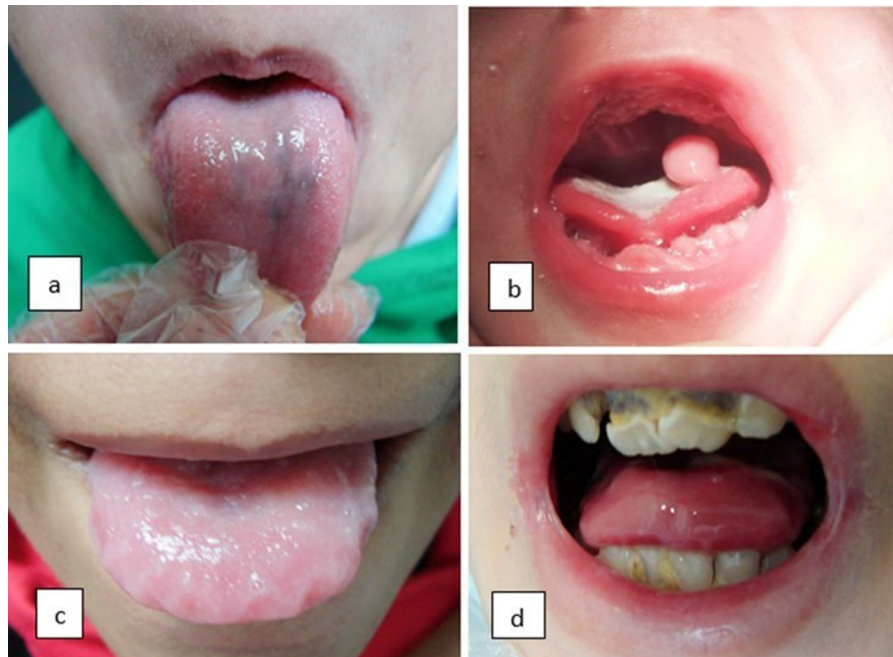


Fig. 2. a) Hemangioma in a patient with Kindler syndrome, b) Fibroma in a patient with oro-facio-digital syndrome, excessive tongue coating and bifid tip of the tongue, c) Limited movement of tongue in a patient with lipoid proteinosis, d) limited movement in a patient with epidermolysis bullosa.



Fig. 3. Spoon tongue in a patient with arthrogyrosis multiplex.

Conclusion

This study described different tongue anomalies seen in a group of patients diagnosed with genetic syndromes, some of these anomalies helped with the diagnosis as the limitation in tongue movement observed in epidermolysis bullosa and lipoid proteinosis patients. Also, a newer classification system for tongue anomalies was proposed, which in our point of view will aid clinicians in diagnosis of genetic disorders associated with such anomalies.

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Conflicts of interest

The authors declare that they have no conflict of interest.

Data availability statement

All available data has been included in the submission.

The data that support the findings of this study are available from the corresponding author upon reasonable request.

Ethics approval

This study received ethical approval from the Medical Research Ethics committee of National Research Center under the protocol number 1424052023.

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