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Hemifacial microsomia: A rare case of a 25 year old female patient with unilateral facial asymmetry (Hemifacial Microsomia) at Al-Abdali hospital, Amman, Jordan

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Abstract
A 25 year old female patient presented to Abdali Hospital in 2019 with features of hemi facial asymmetry, she was refereed to Radiology department from Ear, Nose and Throat department (ENT) for imaging.
The patient underwent Brain and Facial CT scans without IV contrast and also Brain MRI with IV contrast for evaluation, the findings were asymmetrical enlargement and thickening of the left skull bones along with proptosis of the right eye and right mandibular ramus hypoplasia, facial soft tissue swelling associated with right facial muscle wasting.
The patient is medically free with no chronic diseases, the surgical history was also free, and all blood work up was within normal range.
She first noticed facial asymmetry at right side of her face since Childhood, no family history of similar condition among her relatives.
Based on the characteristics of her facial physical examination and imaging finding a diagnosis of Hemifacial Microsomia was made.
The patient was referred back to the ENT referral doctor and conservative therapy was made.
No special procedures like biopsy was made
The patient was asymptomatic at the time of presentation although she reports headaches and decrease hearing at the right side in the past few months.
The most common congenital facial anomaly is cleft palate and lip, HFM is considered the second most common congenital facial anomaly, the incidence is about 1 within in every 3500 to 4000 births, it is a non-progressive nature disease which mean that the affected area at birth will remain similarly affected through growth.
Hence HFM involves both first and second branchial archs the name syndrome of 1st and 2nd branchial archs cames.

Keywords: hemifacial microsomia, facial asymmetry, first and second branchial arch syndrome

Introduction

Background

Hemifacial microsomia was described by Carl Ferdinand von Arlt in Germany during 1881 [1].
It means decrease size of one side or one half of the face, it is believed that it occurs in the early stages of facial development, due to blood supply disturbance to the first and or second branchial arches in the first six to eight weeks of gestation [2].
A 25 year old female patient presented to Abdali Hospital in Amman Jordan, with bilateral facial asymmetry, she was refereed to Radiology department from Ear, Nose and Throat department (ENT) for imaging.
The patient is medically free with no chronic diseases, the surgical history was free, and all blood work up was within normal range.
She demonstrated asymmetry at her right facial half since her childhood, with no family history of similar condition among her relatives.
Based on the characteristics of her facial physical examination and imaging finding a diagnosis of Hemifacial Microsomia was made.
The patient was referred back to the ENT referral doctor and conservative therapy was made.
No special procedures like biopsy was made
The patient was asymptomatic at the time of presentation although she reportsheadaches and decrease hearing at the right side in the past.

Methods

The patient was scanned with Force CT scan in October 2019 and then scanned by contrast enhanced MRI scan using(3 Tesla) machine, preparation of the patient included brief questionnaire about allergic history and state of pregnancy, the CT exam was for Facial Bone without IV contrast, it started by the patient holding her breath and stand still while images was taken, three dimensional (3D) images reconstruction was done, The radiology doctor advised to do Facial MRI, and the patient was given 20 ml

(Dotarem ®) IV contrast at the time of imaging and Brain and Facial MRI was taken.

Discussion

Hemifacial Microsomia (HFM) has many other names such as syndrome of the first and second branchial arch, craniofacial microsomia Otomandibular-dysostosis, lateral facial dysplasia.

HFM results from error during the intrauterine development of the first and second branchial arches, so that is why it affect primarily the mouth, jaw and ear, which results in hypoplasia of these components, usually it is unilateral but both sides of the face are affected in less than 15 % of cases and sometime with asymmetrical involvement.

The most common congenital facial anomaly is cleft palate and lip, HFM is considered the second most common congenital facial anomaly, the incidence is about 1 within in every 3500 to 4000 births, it is a non-progressive nature disease which mean that the affected area at birth will remain similarly affected through growth [3].

HFM lead to underdevelopment and hypoplasia of the mandibular ramus, mandibular joint, muscle of mastications (which consist of the temporalis, masseter, medial and lateral pterygoid muscles), and the ear leading to microtia or even atresia of ear pinna associated with hearing loss due to non-development of the bony and osseous parts of auditory system or absent of both the external auditory canal and system [4].

There is male predilection and right side being more commonly affected that the left side.

Hemifacial Microsomia occurs as a sporadic and it is typically not inherited, in cases of inheritance it may be inherited from one parent, with the pattern of inheritance is mostly autosomal dominant occur when one copy of altered gene in each cell is sufficient to cause the syndrome, in rare cases it occur as autosomal recessive pattern which is occur when the two copies of the gene in each cell have mutation.

The genes involved in Hemifacial microsomia are unknown yet [5].

Extracranial anomalies seen in 55% of patient's inculde other systems like central nervous, skeletal (vertebral), gastroinstinal, genitourinary, and respiratory systems [6].

Goldenhar Syndrome which is a rare congenital condition, should not be confused with HFM, in fact Goldenhar Syndrome include spinal anomalies, lipodermoids and epibulbar dermoids [7].

Treacher Collins syndrome can mimic facial characteristics of hemifacial microsomia in children, the difference between two of them that the Treacher Collins syndrome characterizes by effecting both sides of face while HFM is typically unilateral disease.

During 1969 Dr. Samuel Pruzansky put calssification for HFM:

- **Stage 1:** minimal mandibular hypopalsia
- **Stage 2:** deformed and functional temporo mandibular joint (TMJ) with medial and anterior mandibular condyle dispalcement
- **Stage 3:** non development of the glenoid fossa and mandibular ramus [8].

The other classification system is called the (OMENS) classifications this system founded in 1991 which is more comprehensive, and it is the most commonly used

Table 1: (OMENS) classifications of Hemifacial Microsomia

A. Classification of Pruzansky[21]	
Grade I	Minimum mandibular hypoplasia with all structures present.
Grade II	Condyle, ramus and sigmoid notch are present, but with serious alteration in shape and size.
Grade III	Mandibular ramus can be reduced to a small thin layer of bone, or does not exist.
B. classification of Pruzansky modified by Kaban[22]	
I	Mandibular morphology is normal but small.
IIA	Mandibular ramus is short but of abnormal size; glenoid cavity in right position and functional.
IIB	Glenoid cavity is in an altered position, in inferior, medial, and anterior position.
III	Absence of temporomandibular joint (TMJ)
C. OMENS[23]	
O: Asymmetry of the orbit (Orbit)	
O0	Orbit with normal size and position.
O1	Abnormal orbital size.
O2	Abnormal orbital position (place a position arrow, e.g.: O2 ↑ for upper, O2 ↓ for lower).
O3	Abnormal orbital size and position.
M: Mandibular hypoplasia (Mandible)	
M0	Normal mandible.
M1	The mandible and glenoid fossa are small, with a short ramus.
M2A	The glenoid fossa has an anatomically acceptable position with reference to the opposite TMJ.
M2B	The TMJ is displaced in a lower, medial and anterior way, with a severely hypoplastic condyle.
M3	There is a complete absence of ramus, glenoid fossa and TMJ.
E: Deformity in the outer ear (Ear)	
E0	Normal ear.
E1	Mild hypoplasia, but all structures are present.
E2	Absence of the external auditory canal with variable hypoplasia of the shell.
E3	Lobe poorly positioned, with absence of ear. Lobar remnant is generally moved towards a lower anterior position.
N: Nerve involvement (Nerve)	
N0	There is no facial nerve involvement.
N1	Upper involvement of facial nerve (temporal and zygomatic rami).
N2	Lower involvement of facial nerve (buccal, mandibular and cervical rami).
N3	All rami of facial nerve are affected. Other nerves can be involved, such as trigeminal NV (sensory), hypoglossal N XII, and the rest of cranial nerves with their own numbers.
S: Deficiency in soft tissue (Soft tissue)	
S0	There is no deficiency of soft tissue nor muscle deficiency.
S1	Minimal soft tissue and minimal muscle deficiency.
S2	Moderate – between both extremes, S1 and S3.
S3	Severe soft tissue deficiency due to hypoplasia of subcutaneous tissue and muscle.

Radiological imaging plays critical role in establishing diagnosis and management for HFM, with the CT is the modality of choice with the use of panoramic comparison between the malformed and normal side, CT can give three dimensional imaging (3D) of the facial soft tissue and the underlying bone.

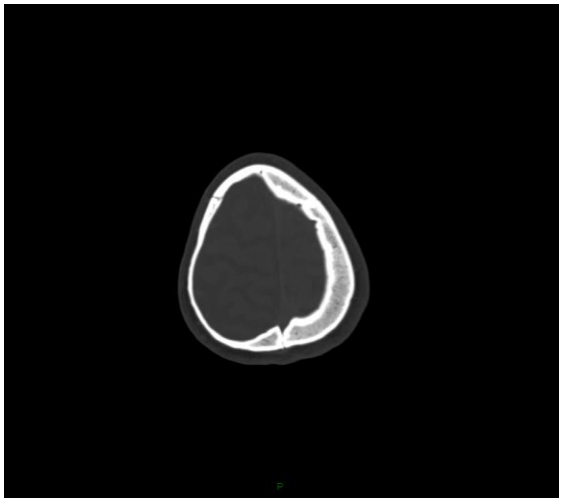

Fig 1: Brain CT scan Bony window showed bone thickening of the left skull bones

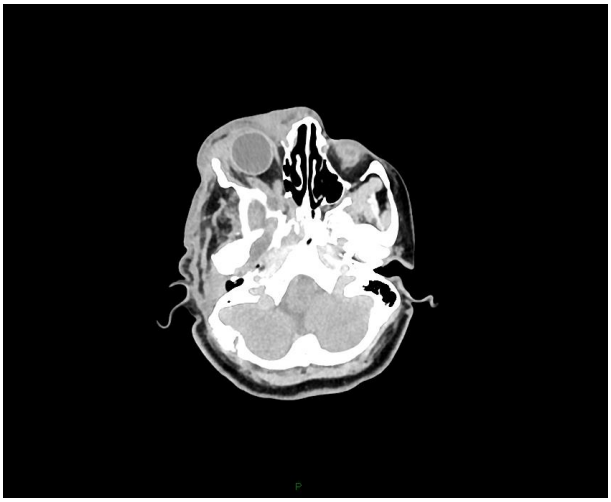

Fig 2: Brain CT soft tissue window without IV contrast shows protusion of the right eye with periorbital soft tissue swelling.

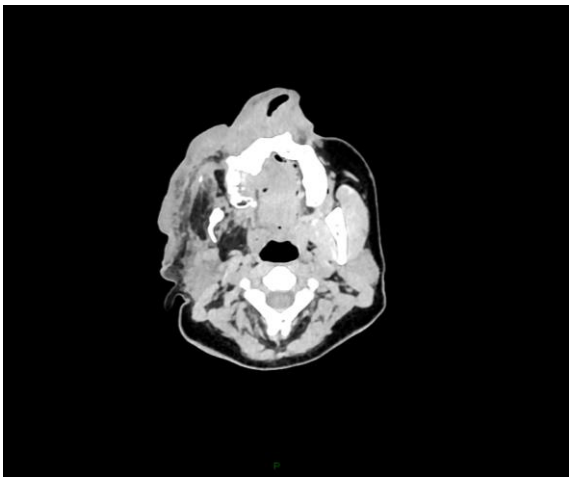

Fig 3: Brain CT scan soft tissue without IV contrast, hypoplastic right mandible ramus with right side facial soft tissue swelling with wasting of the right facial muscles.

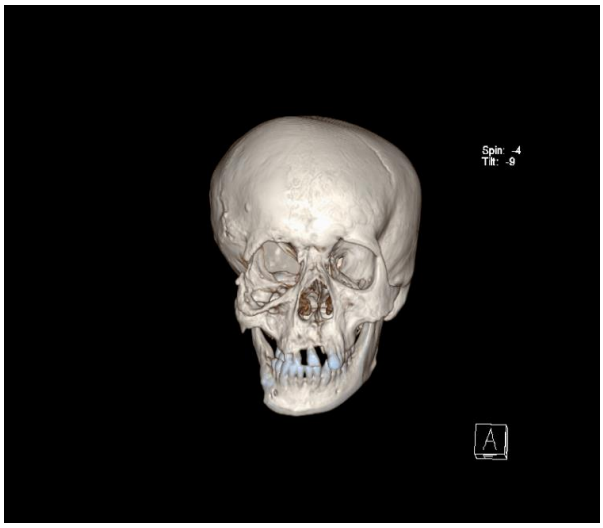

Fig 4: Three dimensional reconstruction for skull shows asymmetry at the right side with hypoplastic right mandiblar ramus and right zygomatic bone, right orbit is lower than left orbit.

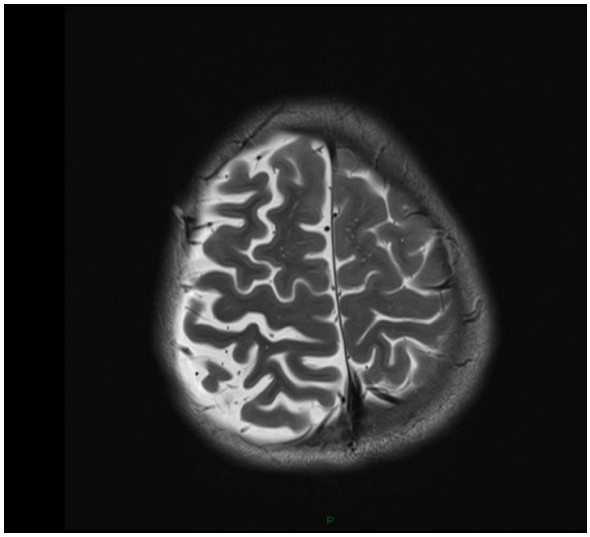

Fig 5: Axial Brian MRI T2 sequence shows right hemisphere brain atrophy.

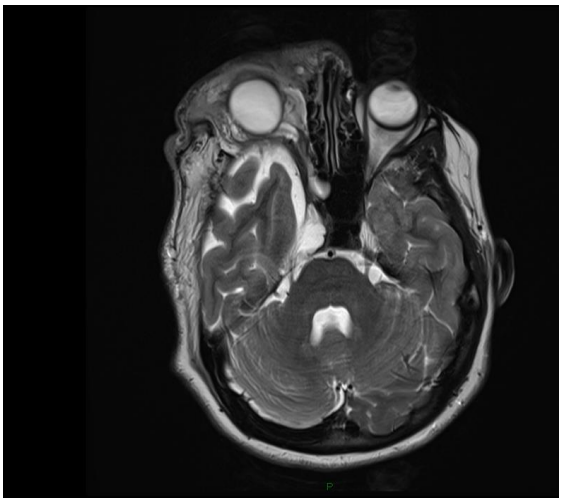

Fig 6: Axial Brain MRI T2 sequence shows proptosis of the right orbit with right peri orbital soft tissue swelling.

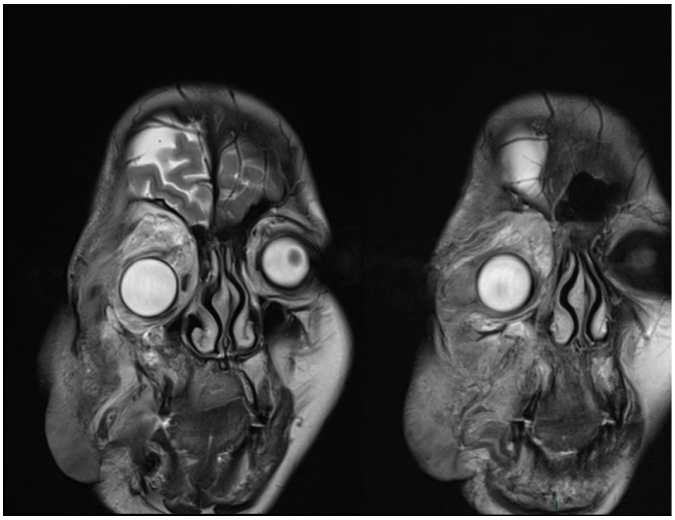

Fig 7: Two Coronal Brain MRI T2 sequence shows right facial asymmetry, with the right eye lower than left eye

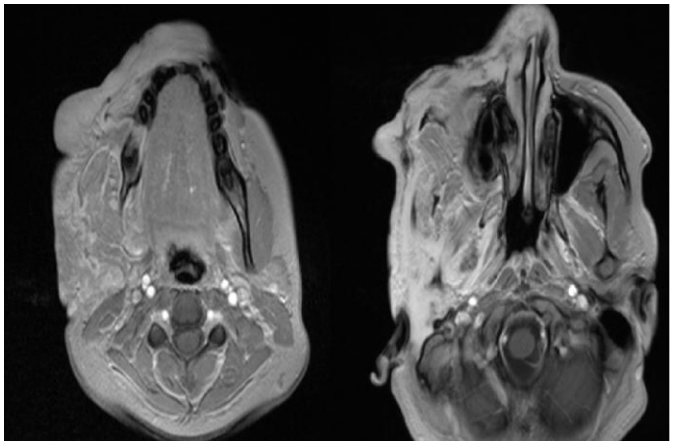

Fig 8: Axial Brain MRI with IV contrast shows right side soft tissue enhancing.

Table 2: (OMENS) classifications for our patient

No	Feature	Our patient
1	Facial Deformity	Mild
2	Orbit Size/Position	Eye in the affected side is lower
3	Absent or small Pinna	Small pinna
4	Pre auricular skin tags	Absent
5	Condylar Aplasia/hypoplasia	Condylar and ramus hypoplasia
6	Malocclusion	Present
7	Impacted/missing teeth	Present
8	Hearing loss	Absent
9	Facial paresis/palsy	Absent
10	OMENS classification	O3M1E1N0S1

Differential diagnosis of Hemifacial microsomia

Patients with Hemifacial microsomia must be distinguished from patients with Goldenhar syndrome, Treacher-Collins syndrome, Traumatic postnatal deformity, Juvenile rheumatoid arthritis, Hemimandibular elongation, Parry-Romberg syndrome, muscle dysfunction, Nager acrofacial dysostosis syndrome, post axial acrofacial dysostosis, Maxillofacial dysostosis and Branchio-oto-renal syndrome [9].

The different between Treacher-Collins syndrome and Hemifacial microsomia is that Tracher-Collins syndrome is always manifested as both zygomatic area with orbital rim deficient, along with absent or deficient condyles muscles and short ramus with retrognatia.

Regarding Goldenhar's syndrome the patient will have ear malformations with the most important difference which is multiple accessory tragi in a mandibular and preauricular distribution with facial asymmetry in 65-75% of cases.

Treatment of hemifacial microsomia is depending on the areas involved, symptoms present and its severity in the patient. In some children with advance cases breathing support or even tracheostomy may be needed after birth especially when the jaw is severely affected, in most cases airway problems can be managed conservatively without need for surgery. Sometimes patients with jaw deformity may need nasogastric tube for gain of weight and growth support. If eye closure is incomplete because of facial paralysis or eye abnormality eye protection or surgery can be performed. Some patients with abnormal ear shape or absent ear may underwent reconstructive surgery to make ear appear normal. Augmentation procedures such as tissue transfer or fat grafting for patients with soft tissue deficiencies can be helpful [10].

Conclusion

Hemifacial Microsomia is non inherited condition in which one side or one half of the face is underdeveloped or hypoplastic and does not grow normally, the most common involved areas are lower jaw, eye, facial nerves, cheek bones and facial muscles may be affected.

It is the second most common congenital facial birth defects after lip and palate clefts, slightly more in male and more to involve right side of the face.

The etiology of Hemifacial in most cases is unknown, and the responsible gene is unknown, it can occurs in people with no family history of hemifacial microsomia and in rare cases it can be inherited.

Ethics approval

This study was approved by ethical committee from our institution and all data was obtained lawfully after taken consent from the patient.

Consent to participate

Efforts to obtain consent form failed because Lack of patient's address, patient is Iraqi national and presence of CORONA Pandemic

Conflict of interest

None.

Funding source

None.

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