CASE REPORT

Cervico-oculo-acoustic (Wildervanck) syndrome: clinicoradiological findings

Farouk Bassam Rihani

Department of Dentistry,
Jordanian Royal Medical
Services, Al-Huson,
Irbid, Jordan

Correspondence to

Dr Farouk Bassam;
Rihani fbrihani@yahoo.com

SUMMARY

We describe a girl presented with facial asymmetry and oral mucosal cleft and with further investigations other anomalies were found including hearing loss, Duane syndrome, Klippel-Feil anomaly, Chiari malformation and accessory bone mass in mandibular ramus leading to the clinical diagnosis of cervico-oculo-acoustic (Wildervanck) syndrome (COAS). The patient underwent surgical occipital decompression by preforming suboccipital craniectomy and C1 posterior laminectomy to relieve the cerebellar tonsillar herniation. Surgical removal of mandibular bony mass was done and the patient is now under orthodontic treatment to correct facial asymmetry and malocclusion.

BACKGROUND

Disturbances in mandibular condylar growth can lead to facial asymmetry. These disturbances have different aetiologies which could be congenital malformations or primary growth disorders or acquired including trauma, and is a main clinical diagnostic marker in many genetic syndromes affecting the first and second branchial arches.¹ Despite the hallmark diagnostic neck features in Klippel-Feil anomaly, facial asymmetry can affect 13% of cases.²

Klippel-Feil anomaly can also be associated with many craniofacial manifestations. These anomalies can include neurological, hearing, ocular, palatal and speech defects.³ Of these syndromes associated with Klippel-Feil is COAS, which is characterised by Duane syndrome and hearing loss. COAS is a rare sporadic syndrome and almost exclusively affecting females with lethality in hemizygous males suggesting an X-linked inheritance pattern.⁴

We present a patient presented with the classical triad of COAS and presented with soft tissue and bony malformations extending from upper cervical vertebra to the oral region and with neurological manifestations.

CASE PRESENTATION

A 9-year-old girl born to consanguineous parents was referred to the paediatric dental clinic for evaluation of facial asymmetry. She had a history of recurrent attacks of dizziness and occipital headaches. After physical examination her height was found to be 119 cm (<3rd centile) indicating proportionate short stature. Her neck was short with low posterior hairline associated with limitation of flexion, extension and lateral rotation neck movements. Craniofacially she had mild facial asymmetry, ocular hypertelorism, deviated nasal septum, low-set ears and mouth breather with incompetent

lips (figure 1A,B). Audiometry revealed moderate sensorineural hearing loss in the right ear. Ophthalmic examination demonstrated decreased visual acuity in the left eye accompanied with type I Duane syndrome in ipsilateral eye. Intraoral examination showed asymmetric mandible with class II malocclusion and anterior open bite. She had limited mouth opening and restricted lateral mandibular movements. On left buccal mucosa there was non-penetrating superficial mucosal cleft extending to left pterygomandibular raphe (figure 2). She had hypernasal speech and her soft palate was immobile with bifid uvula indicating velopharyngeal insufficiency.

INVESTIGATIONS

Lateral cephalogram showed C2–C3 body fusion and C2–C4 spinous processes fusion (figure 3). Orthopantomogram demonstrated short left mandibular ramus, malformed left temporomandibular joint (TMJ) and bony mass in the left coronoid region (figure 4). Brain MRI revealed type I Chiari malformation with anterior displacement of the proximal end of the spinal cord (figure 5A, B). Cranial three-dimensional CT confirmed the presence of an accessory bone mass extending from the left TMJ to the anterior border of ipsilateral coronoid process and malformed left TMJ (figure 6A, B).

Echocardiogram, chest x-ray radiographs, abdominal and renal ultrasounds were within normal limits.

DIFFERENTIAL DIAGNOSIS

Klippel-Feil anomaly can be seen in many syndromes. Differential diagnosis include oculo-auriculo-vertebral spectrum with and without radial defects (OMIM# 141400 and 164210) due to overlapping with COAS

Figure 1 (A) Frontal facial view with hypertelorism and mild facial asymmetry. (B) Lateral facial view with short neck, low posterior hairline and low set ears.

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Figure 2 Intraoral left buccal view with mucosal cleft extending to left pterygomandibular raphe.

in some cases, Mayer-Rokitansky-Kuster-Hauser syndrome (OMIM# 277000) and MURCS (OMIM# 601076, type II Mayer-Rokitansky-Kuster-Hauser syndrome).⁵

TREATMENT

Emergency neurosurgery performed to alleviate the pressure on brain stem by performing suboccipital craniectomy and C1 posterior laminectomy. Surgical excision of accessory bone mass was performed through intraoral incision in left maxillary sulcus to the anterior border of the left ramus. The patient is now undergoing orthodontic treatment to correct her malocclusion and facial asymmetry.

OUTCOME AND FOLLOW-UP

Both surgeries were uneventful. Attacks of headaches and dizziness disappeared and mandibular movements in both opening and lateral excursions improved. Glasses prescribed to improve visual acuity of the affected eye. The patient is now 14 years old

Figure 3 Lateral cephalogram showing fusion of bodies of C2–C3 and fusion of posterior elements of C2–C4.

Figure 4 Orthopantomogram with shortened left ramus, malformed left temporomandibular joint (TMJ) and accessory bone mass in ipsilateral TMJ and coronoid region.

and still under continuous follow-up in neurosurgery and orthodontic clinics.

DISCUSSION

Patients with Klippel-Feil as well as COAS present with many craniofacial and extracranial anomalies which complicate treatment.^{3 6 7} Failure of mesodermal somites segmentation of cervical vertebra during embryogenesis in this patient resulted in Klippel-Feil anomaly with subsequent limitation of normal neck movements and short neck that attributed to her short stature.⁷ Furthermore, fusion of cervical vertebra increase neck rigidity and can complicate anaesthesia induction.⁶

Facial asymmetry in this patient was bony in origin due to malformed left TMJ which restricted the growth of the left side of the mandible compared with the normal growth of the right side of her mandible, and resulted in short left mandibular ramus. This mandibular asymmetry at this age is mild, but also progressive with further growth of non-affected side of mandible and will become marked when patient reaches adulthood.⁷

Occasional abnormal bony masses are seen in COAS patients in the region of mandibular rami as in this case.⁷ This bony mass interfered with normal mandibular movement causing limitation of mouth opening and lateral mandibular movements.

Cleft palate is also seen in some cases with COAS.^{3 6 7} In this patient the oral cleft was in left buccal mucosa extending to pterygomandibular raphe and interfering with normal soft palate movement and causing velopharyngeal insufficiency and hypernasal speech.

Neurologically, different neurological anomalies can occur in COAS. Of high clinical significance is Chiari malformation in which cerebellar tonsils descend through foramen magnum with resultant compression on brain stem.⁶ This patient had the congenital type I Chiari malformation which necessitated emergency neurosurgery to relieve the compression on spinal cord.

Finally, patients with facial asymmetry combined with other cranial and extracranial anomalies need further investigations as this might be life saving as in our patient.

Learning points

- ▶ Cervico-oculo-acoustic syndrome (COAS) and hence Klippel-Feil anomaly are a rare cause of facial asymmetry.
- ▶ Klippel-Feil anomaly might be associated with Chiari malformation which is life-threatening.
- ▶ Accessory mandibular bony masses in COAS can affect normal vertical and lateral mandibular movements.

Figure 5 T1-weighted sagittal (A) and coronal (B) brain MRI with caudal displacement of cerebellar tonsils through foramen magnum and anterior displacement of brain stem.

Figure 6 (A) Lateral cranial three-dimensional (3D) CT scan with accessory bone mass extending from left temporomandibular joint (TMJ) to anterior border of coronoid process. (B) Posteroinferior cranial 3D CT showing malformed left TMJ. Note suboccipital craniectomy and widening foramen magnum.

Competing interests None.

Patient consent Obtained.

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