Congenital Midline Cervical Cleft: A 7 Year Prospective Study

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Abstract Congenital cervical midline cleft (CCMC) is a rare disease of the head and neck region with only a few reported series. The patients attending the ENT or Plastic surgery OPD of a tertiary referral hospital of eastern India were screened for the presence of CCMC. All the patients found to have the developmental abnormality were included in the study and treated. Five patients were identified as having CCMC within the 7 year study duration. Of them three were female and two male. The youngest patient was a 2 day old baby and 25 years was the maximum age of presentation in the present series. All except the new born were treated by surgical excision and primary repair of the neck defect. In literature CCMC has been described almost exclusively in the Caucasian population. The present series documents the occurrence of this developmental abnormality in the Indian population. This is perhaps the first case series on CCMC to be presented from the Indian subcontinent.

Keywords Congenital cleft · Cervical · Midline · Surgical excision

Introduction

Midline cervical cleft is a rare congenital abnormality of the neck presumably caused due to failure of fusion of branchial arch derivatives in the intra-uterine period of life. Less than 100 cases have been published in literature so far after being first described by Bailey in 1924 [1]. PubMed search with the MeSH terms of congenital, cervical, midline and cleft shows 83 articles on this topic but mostly a report of two or three cases. In this article a case series of 5 patients having congenital cervical cleft is being reported with their clinical presentation and management and a review of literature on this rare disease.

Materials and Methods

- (i) Study population: patients presenting to the ENT or Plastic surgery OPD of a tertiary referral hospital in Kolkata, India.
- (ii) Study duration: May, 2005 to September, 2012.
- (iii) Study design: retrospective case series.
- (iv) Selection criteria: patients of any age presenting with a midline cervical cleft was included in the series irrespective of the presence or absence of other otolaryngological or systemic disease.
- (v) Study protocol: detailed history was taken for each of the selected case followed by extensive clinical examination to detect the presence of any other abnormality. It was further supplemented with other investigations wherever necessary. Management of

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each of the patients was individualized according to the extent and nature of the cleft and the excised specimen was histologically examined. The patients were kept under regular follow up after discharge.

Results

Total five patients of congenital midline cervical cleft were identified and documented during the period of study. The youngest patient was a newborn male having a complete cleft up to the sternum (Fig. 1). Out of total five cases, three were female (Figs. 2, 3) and two were male. Four cases were surgically treated by excision along with Z-plasty (Fig. 4). The newborn patient was undergoing staged reconstruction at the time of writing this report (Table 1).

Discussion

Cleft lip or palate is very frequently encountered in clinical practice, but midline cervical cleft is a very unusual disease. Tessier in 1973 presented a classification of cranio-facial cleft according to which, the midline cervical clefts was a variant (caudal extension) of the number 30 cleft. This group includes mandibular process cleft, branchio-genic medial axial B2 cleft and midline branchiogenic syndrome [2].

Cranio-facial clefts usually occur due failure of fusion between the facial processes derived from the first and the second branchial arches during the third–fourth weeks of intra-uterine life. The severity of cleft depends upon the extent of mesoderm proliferation within the cleft which occurs as a natural healing effort to close the gap formed due to the failure of fusion of the branchial arch derivatives

Fig. 2 A 19 year old patient showing all components of a midline cervical cleft

Fig. 3 Prominent hood of skin in a midline cervical cleft

Fig. 1 New born baby having a complete cleft extending from the tongue up to the sternum

Fig. 4 Per-operative photograph showing the cleft being excised

Table 1 Collated data of the patients included in the present series

Serial	Age	Sex	Clinical presentation	Location of the sinus	Associated abnormality	Remarks
1	3 years	Female	Discharging midline sinus present since birth with a hood of skin in its superior aspect	Between cricoid and supra-sternal notch	None	Treated by surgical excision
2	18 years	Male	Discharging scar tissue in midline of neck with limited neck mobility	Subcutaneous cord like structure in midline starting from the scar	Congenital otosclerosis	Release of scar contracture, excision of the cord like tract followed by stapedotomy
3	New born	Male	Midline cleft from mandible up to sternum	Complete midline cleft	Bifid tongue and the soft tissue of the floor of oral cavity, hypoplastic/absent strap muscles of neck.	Reconstructive surgery along with plastic and paediatric surgeons.
4	25 years	Female	Midline cleft with a superiorly located hood of skin	Extending from superior border of thyroid cartilage up to the suprasternal notch	None	Surgical excision with Z-plasty
5	19 years	Female	Midline cleft with a prominent hood	Extending from superior border of thyroid cartilage up to the cricoid	None	Surgical excision with Z-plasty

Reference [2]

in the early embryonic period [2]. This theory of incomplete fusion of branchial arches also explains the spectrum of symptoms that these patients may present with, ranging from a simple cord like structure in the neck to a extensive midline defect from the mandible up to the sternum of a new born child as evidenced in the present series (Fig. 1). However over the years various other theories of origin has been proposed by several authors like improper interaction between ectoderm and endoderm, fault in the differentiation of the mesenchymal tissue, from thyroglossal remnant by exteriorization, amniotic adhesions, pericardial roof producing increased pressure on the cervical region of the developing embryo and so on. But the theory of incomplete fusion of the branchial arches seems to explain the disease more rationally per se and hence has been the preferred etiological hypothesis of authors worldwide. Deficient or delayed migration along the second branchial arch is the commonest causative factor which causes the isolated midline cervical clefts with minimal deformity. Incomplete fusion of the second arch derivatives with the first and/or the third arch produces more extensive anomalies as the case may be [3]. A recent study of the genetics of this disease was done in 2012 in three cases but without any confirmatory findings [4].

This entity is more commonly found in the female though not statistically validated yet in any literature. Caucasian females are widely assumed to be the commonest race affected but the malformation is also common in Indian population as evidenced by the present series.

Presentation is usually at birth (Fig. 1), but very frequently the patients seek medical opinion later in life particularly in third world countries like India (Figs. 2, 3). Almost all the patients have a midline defect in the neck that can be situated anywhere between the lower lip and the sternal notch. The lesion is classically described to consist of three parts—a superiorly located nipple like projection formed by a hood of skin, middle fibrous or chord usually covered by reddened epithelium and a inferiorly located sinus that runs for varying distance (Fig. 3). There may be discharge from the sinus; however it is generally scanty and serous in nature at birth which ceases by itself after a few months [5]. The source of the discharge is assumed to be from the presence of heterotopic salivary tissue inside the sinus tract. The fibrotic chord may cause contracture of the neck muscles and torticollis which is the presenting symptom in some patients [5]. In extreme cases there may be complete cleft formation extending from the lip up to the sternum along with hypoplasia of mandible, hypoplasia/aplasia of the hyoid bone and the thyroid cartilage. There may be associated cleft lip, bifid tongue and even bifurcated sternum along with an associated delay in mandibular development and hypoplasia or absence of neck structures such as the hyoid bone [1]. The third case

of the present series presented with such extensive pathology. Co-existing developmental anomalies like thyroglossal duct pathologies or branchial cysts has been described by various authors [3, 6]. No association with otosclerosis has been detected yet though it was present in one of our cases. But deafness due to heterozygous mutation has been reported by [7].

Diagnosis of this entity is based solely on proper history and clinical examination. Ultrasound examination or radiographic examination with dye infusion is done only when a co-existent pathology is suspected like a thyroglossal cyst anomaly or a branchiogenic cyst.

Opinion varies as regards to the to the histological characteristics of CCMC; according to Bergevin et al. [8] the CCMC remains covered by an ectodermal or endodermal mucosal area and not by epidermis. But in their series of five cases, Derbez et al. [9] reported that the CCMC was covered by respiratory type of epithelium in all the patients. However more recent opinion considers stratified squamous epithelium having surface parakeratosis with absent adnexa in the dermis to be the pathognomonic histological characteristic of congenital cervical midline cleft [10]. In the present series the excised tissue histologically presented the characters of stratified squamous epithelium and absence of adnexal tissue in all the four operated cases.

Treatment of congenital cervical cleft is entirely surgical for aesthetic purposes, to prevent cervical contractures and to cure other associated congenital abnormalities. The defect should be repaired as soon as possible and preferably not by simple surgical closure. The preferred procedure is excision of the cleft along with the sinus and repair by multiple Z plasty techniques as advocated by Gargan et al. [11] in their pioneering study of 12 cases spanning 30 years. However more advanced cases having hypoplasia of mandible, absent hyoid and/or thyroid cartilage or other supporting structures of the neck warrants extensive teamwork between plastic surgeon, head & neck surgeon,

facio-maxillary surgeon supported by psychologists, speech therapist, paediatrician and very dedicated nursing care [12]. These cases require multiple surgical procedures to be able to lead a meaningful life.

Conflict of interest The authors declare that they have no conflict of interest.

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