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Original Article

Congenital Anomalies Presenting as Head-Neck Swellings: A Study of 50 Cases

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Abstract:

Purpose*: To analyse the prevalence, distribution and presentation of different congenital head-neck lesions with their age, sex, site and side predilection.*

Methods*: 50 patients of head-neck congenital swelling was selected from january/2013 upto december/2013 in the OPD of otorhino-laryngology, head and neck deptt. of Dhaka medical college hospital under a specific prospective study protocol.*

Results*: The most frequent swelling was thyroglossal cyst(42%), followed by branchial arch anomalies (18%),dermoid cyst(14%),pre-auricular sinus(14%),lymphangioma(8%) and haemangioma(4%).Amongst the cervical swellings the sequential preponderence was thyroglossal cyst(52%),branchial arch anomalies(23%),dermoid cyst(13%), lymphangioma(10%) and haemangioma (2%).The majority of branchial arch anomalies(100%) were of second arch. The majority of patients were of first(52%) and second(30%) decade and the male to female ratio for thyroglossal cysts was 1:1.1.The majority of lesions were painless swelling and all were surgically excised.*

Conclusion*: The overall frequency and age-sex distribution of congenital head-neck swellings as well as site, nature and type specific predominance of some of them in OPD of Dhaka medical college hospital are almost similar to international findings.*

Introduction:

Head-Neck swellings in children and adolescents comprise a separate diagnostic and therapeutic category, and they can be broadly divided into congenital and inflammatory/infective groups since neoplastic head-neck swellings are rare in this age group.[1] So, congenital swellings like cyst, sinus and fistula must be considered in the diagnostics of head-neck masses in children and young adults.[2] In children, common neck swellings are nodal swelling (suppurative/ nonsuppurative), congenital cyst (may present becoming infected) and neurogenic tumours.[3] For masses over 2 cm in diameter in patients over 35 years of age, 80% of nonthyroid neck masses are neoplastic of which 80% are metastatic and 75% are with primary sites above the clavicle but in children 90% neck masses are benign conditions of which 55% are congenital.[4,5] The congenital swellings are, in descending order of frequency, thyroglossal duct cyst, branchial arch anomalies,cervico- facial dermoids, preauricular sinuses, lymphangiomas and haemangiomas; and rarely,cervicofacial vascular anomalies, thymic anomalies, laryngocele,laryngeal cyst and bronchogenic cyst etc.[6-15]

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Thyroglossal duct anomalies are the second most common paediatric neck swelling only next to adenopathy and they occur in approximately 7% of population, although only a few of these is ever symptomatic.[2,16] Thyroglossal cyst is the most common cyst, upto 50% present in early adulthood with over 30% presenting before 10 years of age.[3] Most cysts present as midline masses of 2-3cm in diameter just subhyoid/adjacent to hyoid(66%), 25% as suprahyoid and just over 10% being related to the thyroid; they may present as sinuses resulting from spontaneous rupture or surgical drainage or with concurrent or prior infection but most commonly as painless swelling. [3,17] They may be at any level from foramen cecum to superior mediastinum, 90% in midline but 10% on either side with 95% being on the left, mean age of presentation being 5 years ranging from 4 months to old age and rarely thyroglossal duct carcinoma may be the presenting entity,they are usually sporadic but rarely may be a familial variety of autosomal dominant in prepubertal girls.[18] Two thirds of thyroglossal duct anomalies are diagnosed within the first 3 decades of life,with more than half being before age 10 years mostly in toddlers and older children with midline swelling moving with tongue protrusion and swallowing and about 10% arise as submental swellings confusing with dermoid cysts-their congenital origin is only suspected when recurrent or persistent drainage follows surgical excision or spontaneous rupture of an abscess formed in the cyst,this drainage can occur through foramen cecum resulting fetor oris.[1,2] Branchial cysts, sinuses and fistulae compose approximately 30% of congenital neck swellings.[2] and they represent about 20 % of cervical masses in children.[18,19] Strangely, although most remnants have usually been present since birth, branchial cysts most commonly present in adolescence and early adulthood,typically between 10 and 40-years-old, although they can occasionally present at any age[1,3,7] and they got no sex preponderance but with slight left sided predominance excepting the third and fourth arch anomalies are almost exclusively common on the left;[3,9,10] and they are bilateral only in about 1% of the cases.[18,20] The relevant branchial arch can be identified from the distinct anatomical site of the anomalies, the second arch cyst and sinus at the anterior border of the upper and middle third respectively of the sternocleidomastoid muscle and the first around the auricle and the third and fourth at more lower level of the muscle-thus they can be in parotid,neck and mediastinum.[2,3,7] 60% of branchial cysts are in the upper third of the neck, although may be anywhere of neck, and parotid gland;80% present as persistent and 20% as intermittent swelling,70% are clinically cystic although upto 30% may be solid and in upto 40% cases,patients may had URTI prior to noticing the mass[18];the cysts may confuse with reactive lymphadenopathy or infected dermoid cyst or childhood rhabdomyo-sarcoma or lymphoma, tuberculoma and neurofibroma in young adults or metastatic node in patient over 35 years of age.[3] Second branchial arch anomalies represent 95% of all branchial apparatus anomalies while the first arch only 1% -less than 5% according to another series, third and fourth arch rarely.[2,21] Branchial cysts usually appear fairly quickly in relation to an upper respiratory infection, as oval masses of 4-6 cm in diameter, often tender, the swelling may be intermittent and occasionally it is overtly infected with overlying erythema and pain at

presentation.[3] Branchial fistulae or sinuses usually present in infants or young children,usually during the first decade, as weeping defects/sinuses with or without internal opening but occasionally as infected swellings around the sinuses along the anterior border of the muscle at the junction of the middle third and the lower third.[3]

The preauricular sinus also variably termed as preauricular pit or preauricular fistula or preauricular tract or preauricular cyst is not uncommon of which prevalence in USA is 0.1-0.9%, in England 0.9%, in Taiwan 2.5%, in some areas of Africa 4-10%.[22,23] and in mostly it is noted during routine ear,nose and throat examination, though it can present as an infected and discharging swollen sinus.[8] Over 50% cases, they are unilateral, sporadic,commonly with right sided predominance and when bilateral likely to be inherited(autosomal dominant which determines the branchio-oto -renal syndrome).[24,25] The sinus is usually located at the anterior margin of the ascending limb of the helix , it is also located along the posterosuperior margin of the helix, the tragus or the lobule.[26,27] The Preauricular sinus is apparent at birth,may be some sebaceous discharge and can be left safely excepting when recurrent episodic inflammation leads to abscess formation-since lined by squamous epithelium,spontaneous resolution is unlikely.[21] 20% of all dermoid cysts are found in neck and 30% of these in the face; they make up 28% of all midline cysts, with no sex predominance.[18] Cervical dermoids represent only 20% of head-neck dermoids[16], they can present at any age, but majority present in patients aged under 6 years with no sex predominance, often before 3 years[3,16] while another series declares the peak age of its incidence being usually second or third decades and usual site of presentation being submental region, above or below the mylohyoid muscle, sometimes as an inflammed swelling[18.] The one-third lymphangiomas are found in the oral cavity and cheek, and more than a quarter in the neck.[3] Lymphangiomas occur often in posterior triangle where tissue planes are looser than in lips, tongue and cheek. As these enlarge, they may involve cheek, parotid, oral cavity, mediastinum or axilla.[3] Majority of lymphangiomas are present at birth and may now be diagnosed prenatally, their presentation after 2 years of age is extremely rare but their recurrence may be after many years of previous treatment; sometimes they can manifest for the first time in young adults and they can present anywhere in head and neck.[3] They are usually asymptomatic but the cosmetic distress. They feel cystic and transilluminate. Another vascular swellings, the haemangioma and vascular malformation, are not uncommon in children and some of them present, often, as birthmarks.

Materials and Methods

Type of study: prospective study,50 cases, random selection.

Place of study: Department of ENT, Dhaka medical college hospital.

Period of study: From january/2013 to december/2013.

Number of patients: 50 patients of head-neck swellings were selected randomly.

Inclusion criteria: all congenital head-neck swellings were selected.

Exclusion criteria: malignancy, inflammatory swellings and other thyroid swellings were excluded.

Follow up: done with standard protocol.

Results:

Table - I

Site distribution of the head-neck congenital anomalies(n=50)

site	pre-auricular sinus	thyroglossal duct cyst	branchial apparatus cyst/sinus	dremoid cyst	Lymph-angioma	Haem-angioma
Head	7	-	-	2	-	1
Neck	-	21	9	5	4	1
total	7(14%)	21(42%)	9(18%)	7(14%)	4(8%)	2(4%)

Table-II

Distribution of the cervical anomalies(n=40)

Lesions	No. of the patients	Percentage (%)
Thyroglossal cyst	21	52%
Branchial cyst/sinus	09	23%
Dermoid cyst	05	13%
lymphangioma	04	10%
haemangioma	01	2%

Table-III

Age distribution of the patients(n=50),as a whole

age(years)	No. of the patients	Percentage
0-10	26	52%
11-20	15	30%
21-30	07	14%
31-40	02	04%

Table - IV

Age distribution of the patients(n=50),different congenital anomalies wise

age (years)	no. of the patients of thyroglossal cysts,n=21	no. of the patients of branchial arch anomalies,n=09	no. of the patients of preauricular sinus,n=07	no. of the patients of dermoid cysts,n=07	no. of the patients of lymphan-giomas,n=04	no. of the patients of haeman-giomas,n=02
0-10	07(33%)	03(33%)	04(57%)	0-3yrs=03(43%)	at birth=02(50%)	at birth=01(50%)
11-20	10(48%)	02(22%)	02(29%)	4-6yrs=02(29%)	0-2yrs =01(25%)	0-5yrs=01(50%)
21-30	03(14%)	03(33%)	01(14%)	7-10yrs=01(14%)	3-5yrs=01(25%)	
31-40	01(5%)	01(12%)		11-15yrs=01(14%)		

Table - V

Sex distribution of the patients of thyroglossal cysts,branchial arch anomalies and preauricular sinuses(n=37)

sex	no. of the patients of thyroglossalcysts, n=21	no. of the patients of branchialarchanomalies, n=09	no. of the patients of preauricularsinuses, n=07
Male	10	05	03
Female	11	04	04

Table-VI
Site predominance of the thyroglossal cysts/sinuses(n=21)

Site	No. of the patients	Percentage
Suprahyoid	05	19%
adjacent to hyoid	13	62%
Thyroid related	02	9%

Table-VII
Side predilection of thyroglossal cysts(n=21)

Site	No.of the patients
midline	19
left paramedian	02

Table-VIII
Presenting status of the thyroglossal duct cysts/sinuses(n=21)

Status	No. of the patients	Percentage
Painless cysts	11	52%
Sinuses	04	19%
Concurrent or prior infection	06	29%

Table-IX
Surgical status of the thyroglossal cysts(n=21)

status	No. of the patients	Percentage
painless swelling	12	57%
infected	03	14%
abscess	02	10%
operated abscess	03	14%
sinus tract operation	01	5%

Table-X
Site distribution of the branchial arch anomalies(n=09)

Site	Arch	Cyst/sinus	No. of the patients	Percentage
sub-aural	first	-	-	0%
upper third of SCM	second	cyst	04	44%
lower third of SCM	second	sinus	05	56%
Lower neck	third/fourth	-	-	0%

Table -XI
Side predilection of the patient of branchial arch anomalies(n=09)

Side	no. of the patients	ratio
Left	05	5:4
Right	04	

Table -XII
Side predilection of the patient of pre-auricular sinus(n=07)

side	no. of the patients
Unilateral	05(right-03,left-02;right:left=3:2)
Bilateral	02

Discussion:

Thyroglossal duct cysts (sinuses/fistulae) are about 50% of all congenital neck swellings[3]. In our study,thyroglossal cysts are 42% of total head-neck and 52% of total neck swellings;33% presented in first and 48% in second decade;male:female ratio is 1:1.1;62% adjacent to hyoid ,19% suprahyoid and 9% related to thyroid-2 out of 21 were on the left with remaining in the midline; 52% presented as painless swelling, 29% with concurrent or prior infection and 19% with sinuses and14% was with only infection, 10% with abscess,14% underwent abscess operation and 4% underwent sinus tract operation (Sistrunk procedure)-all the results resemble,more or less,the other accepted studies[3,6] sinceTaiseer Hussain Al-Khateeb et al reported thyroglossal cysts being 53% of cervical masses and Michael Gleeson et al reported above 30% presents before 10 yrs and upto 50% in adult; male:female=1:1;65% just subhyoid,25% prehyoid and 10% related to thyroid but they reprortd that most cysts present with sinues whereas John C Watkinson et al reported that 15% present with sinuses which is near to our result. Thyroglossal duct cysts are usually lined by columnar epithelium with small glands frequently containing thyroid colloid[3]- a solid mass indicating median ectopic thyroid, occurs only in 1% to 2% cases, or increased TSH should herald thyroid scanning[17,28] but we performed scanning in each case even though no solid mass or increased TSH were found; USG guided FNAC and CT/MRI may be needed to exclude malignancy in large cyst since although rare, thyroglossal duct carcinoma may be the presenting entity[18] these tools are also needed to exclude lingual thyroid. Excision of a core of tissue upto foramen ceacum (Shalang procedure) should be practised during the first procedure to prevent recurrence[17,29].

In our study, branchial cysts/sinuses were 18% of total head-neck and 23% of total neck swellings, 56% were sinuses and 44% cysts, about 50% were presented during childhood,100% were of second arch origin, male:female=1:0.8 and got slight left predominance(5:4)-the results mimic, more or less, other accepted studies[1,2,3,6,7] since Taiseer Hussain Al-Khateeb et al reported branchial arch anomalies being 22% of neck swellings and Stephanie P. Acierno et al reported them being presented usually in childhood and early adulthood with male female equal prevalence but second arch comprises 95% of all branchial arch anomalies whereas it is 100% in our study-Michael Gleeson et al reported that the anomalies got slight left sided predominance.Branchiogenic carcinoma are rare and it is only 0.3% of all head-neck carcinoma[30], distinguishing between a primary lesion arising within an anomali and a metastatic one from an occult primary is difficult[16]; USG guided FNAC rules out malignancy or clarify the diagnosis in adults-this clarification is not necessary in children and incisional biopsy should not be performed because of more difficult resection[19]; FNAB when lymphnodes are necrosed as is seen in SCC or TB and sonogragrphy fails to distinguish them from an infected cyst-CT/MRI(CT is the current study of choice) helps then, CT fistulogram delineates the tract upto 64% helping the surgical plan to prevent recurrence, virtually finally the tract should be ligated before its avusion from tonsillar fossa to prevent recurrence[31,32] and finally Barium esophagogram with a 50% to 80% sensitivity helps for third and fourth arch fistulae[43]..In our study, preauricular sinuses were 14% of congenital head-neck anomalies,57% presented during the first decade,71% were unilateral with few 3:2 right predominance-resembling other accepted results[24,25] since L.J. Paulozzi et al reported

over 50% cases are unilateral with right sided predominance. The majority cases were asymptomatic, isolated asymptomatic case requires no treatment. Preauricular sinus may lead to subcutaneous cyst formation which can be intimately related to tragus or anterior crus of helix; since the sinus tract may vary in length, branch and course a wide local excision under magnified visualisation is essential to prevent recurrence[5,24].

Dermoid cysts may occur from implantation secondary to puncture wounds, and they can be close to hyoid and move with swallowing or tongue protrusion leading to confusion with thyroglossal duct cyst. These dermoids usually originate from ectoderm and mesoderm and head-neck is the most common site of their occurence,they present predominately in the midline,may be lateral to submandibular gland[2,3]. In our study,dermoid cysts are 14% of congenital head-neck anomalies and 13% of cervical congenital anomalies and 72% were presented by the age of 6 years-the results mimic the other accepted ones[3,6,16] since Taiseer Hussain Al-Khateeb et al reported dermoid cysts are 11% of cervical masses, Michael Gleeson et al reported that majority present before 6 yrs and Enepekides DJ reported they are often diagnosed before 3. Infection is rare but the cysts can rupture due to gradual accumulation of sebum and present with granulomatous inflammation-they are lined by epithelium and contain epithelial appendages like hair,hair follicles,sebaceous glands,sweat glands differentiating them from epidermoid cysts[17]; rarely, there may be a communication through the calvarium and two dermoid elements occurring on either side of the bone resembling a dumb-bell tumour[1]. Physical examination is sufficient to diagnose the cysts but USG helps to delineate the depth and relation with hyoid bone but when the cyst is inflammed FNAC is helpful to distinguish between a ruptured dermoid cyst and an infected thyroglossal duct cyst[2] and a Sistrunk procedure should be perfomed if it is attached to the hyoid bone to prevent inadequate excision of an atypical thyroglossal duct cyst[33]. Lymphangiomas are degenerative lesions arising from lymphatics and only the cavernous type composed of dilated lymphatic spaces,often with fibrous adventatia and presenting as painless diffuse swelling represents 30-40% or 40% of all lymphangiomas[3,18];small vesicle like lesion thin-walled capillary lymphangiomas require no treatment and another life threatening type cystic hygroma composed of flat endothelium lined cysts and sinuses,communicated or isolated,either grows progressively or remains static, but with intralesional bleeding or infection it imparts rapid swelling,severe pain even progressive airway compression-airway problem may be without bleeding or infection for the largest cystic hygroma[34]. In our study,lymphangiomas were 8% of all congenital head-neck anomalies, 10% of all cervical congenital anomalies,100% were presented by 2 years of age and 1 out of 4 was a recurrent case-results are more or less similar to other studies[3,6,19] since John C Watkinson et al reported that lymphangiomas are usually present at birth but Taiseer Hussain Al-Khateeb et al reported lymphangiomas are 6% of cervical masses which is lower than our result. Over 60% cystic hygromas are in the neck and these multicystic mass infiltrated in the tissue plane never regresses spontaneously[1]; so meticulous wide surgical resection is essential to prevent recurrence,a variety of sclerosing agents are used(the latest one is(FK432) for the recurrent cases.Antenatal diagnosis by USG is possible for larger masses; affected infant should be managed in a higher centre[21]. Capillary

haemangioma presents as birth marks which disappears over time usually, arterial (plexiform) haemangioma is pulsatile and venous haemangioma is most versatile. In our study,haemangiomas were 4% of all head-neck swellings and 2% of all cervical swellings; all presented by 5 years of age-the result of prevalence rate differ,a bit, with some accepted studies[6] since Taiseer Hussain Al-Khateeb et al reported haemangiomas are 7% of cervical masses which is higher than our result but .These haemangiomas require FNAC,CT scan and even sometime angiogram for their confirm diagnosis and extension realisation-meticulous extensive resection is essential. Patients been diagnosed in OPD, underwent surgery in IPD and then followed up in OPD imparting some lacking in concrete correspondence is the limitation of our study.

Conclusion:

A thorough understanding of the embryology and anatomy of each of the congenital lesions is necessary to provide accurate diagnosis and appropriate surgical therapy which are essential to prevent recurrence. Any recurrent case, if any unlikely, should be and must be handled in higher centre.

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