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· 论著 ·

先天性颈正中裂 1 例并文献复习

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摘要:目的 探究先天性颈正中裂(congenital midline cervical cleft, CMCC)的发病特点及胚胎学病因,并讨论其手术治疗方案。**方法** 明确诊断后,全麻行"颈前正中裂切除术+双 Z 形皮瓣修复术"。回顾总结 1 例 CMCC 患者的临床资料并文献复习。**结果** 本例患者术后颈部切口一期愈合,随访 1 年无复发。**结论** CMCC 是一种极为罕见的颈部发育畸形,其胚胎成因可能与第一/二鳃弓在中线的闭合不全有关。手术完整切除并一期整复,是首选的治疗手段。

关键词:先天性畸形;鳃裂畸形;颈正中裂;双 Z-成形术
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Congenital midline cervical cleft: a case report and literature review

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Abstract: Objective To explore the characteristics, therapy, and embryological etiology of congenital midline cervical cleft (CMCC). **Methods** After pathological diagnosis revealed a skin fistula, the patient underwent surgical treatment: resection of a contractile, longitudinal neck sinus, skin tag, skin lesion and followed by double Z-plasty. And the clinical data of a case of CMCC was retrospectively reviewed along with a review of related literature. **Results** The postoperative neck incision of the patient healed in one stage. No evidence of recurrence was found during the 1-year follow-up. **Conclusion** CMCC is a rare congenital abnormality of the neck. CMCC is thought to occur through a failure of the first or second branchial arch fusion. Double Z-plasty is a common technique for the prevention of contracture and restoration of the anterior contour of the neck.

Key words: Congenital abnormalities; Branchial cleft abnormalities; Midline cervical cleft; Double Z-plasty

先天性颈正中裂(congenital midline cervical cleft, CMCC)是一种临床极为罕见的颈部发育畸形[1-2],其发病率占颈部先天性畸形的 1.7% ~ 2.0%[3]。据国外文献统计,截止 2015 年仅记载 205 例,其中男 66 例、女 79 例,60 例未标明性别,我国发病 4 例[4-6]。手术完整切除并一期整复是首选的治疗手段。

本文总结分析耳鼻咽喉头颈外科 2020 年收治的 1 例先天性颈正中裂患者的临床特点,综合文献总结如下。

1 资料与方法

1.1 一般资料

患者女,26 岁,生后下颈皮肤凹陷及肉赘伴溢液 26 年,曾在外院被诊为第二鳃裂瘘管,未予处理。2020 年 12 月 8 日就诊于广东省人民医院耳鼻咽喉头颈外科。查体:颈前胸骨上窝中线处皮赘(见图 1A),皮肤凹陷、潮红,范围约 3.5 cm×2.0 cm,伴上方肉赘,长径约 1.5 cm(见图 1B),稍有颈伸受限,头颈部皮肤未见其他瘘口。颈部磁共振成像(magnetic resonance imaging, MRI)示胸骨上方中线处皮肤见一异常信号影,内侧缘可见一长约 15.0 mm 的管状异常信号影与皮下脂肪相通(见图 1C、图 1D)。无相关先天疾病家族史。入院诊断:先天性颈正中裂。

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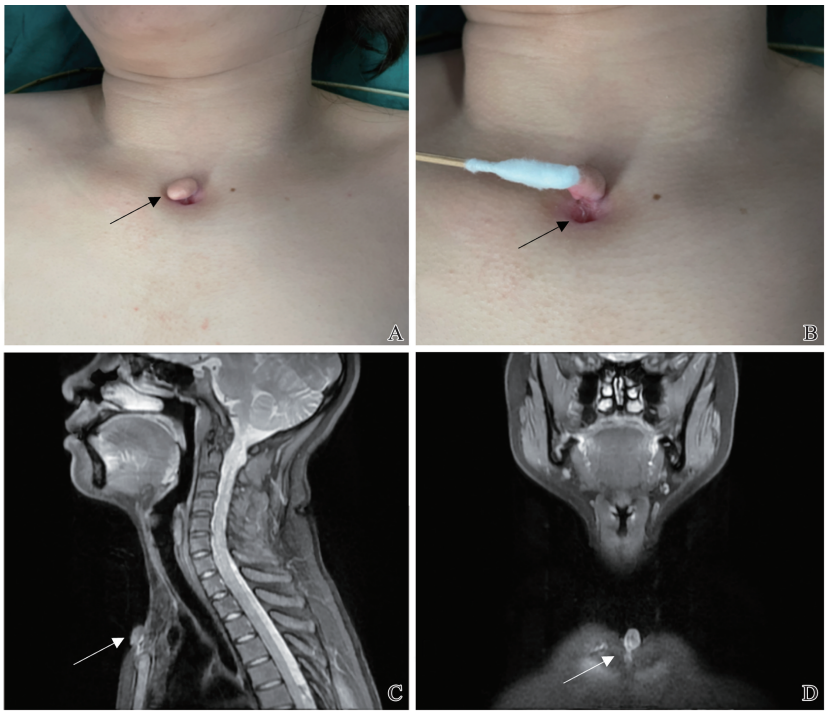

图 1　病损外观(A-B)及影像(C-D)
Figure 1　The appearance (A-B) and imaging (C-D) of lesions

1.2　治疗

2020 年 12 月 10 日,患者全麻下行“颈前正中裂切除术+双 Z 形皮瓣修复术”(见图 2A)。设计下颈前纵向梭形切口,涵盖皮肤凹陷及皮赘,探查窦道长约 2 cm,盲端深至胸骨柄浅面(见图 2B、图 2C)。病灶切除后,设计双 Z 形皮瓣(见图 2D),修整、错位双层间断缝合(见图 2E)。术后 1 个月,颈部伤口愈合佳,无瘢痕(图 2F)。术后病理符合皮肤瘘管(图 3A、图 3B)。随访 1 年无复发。

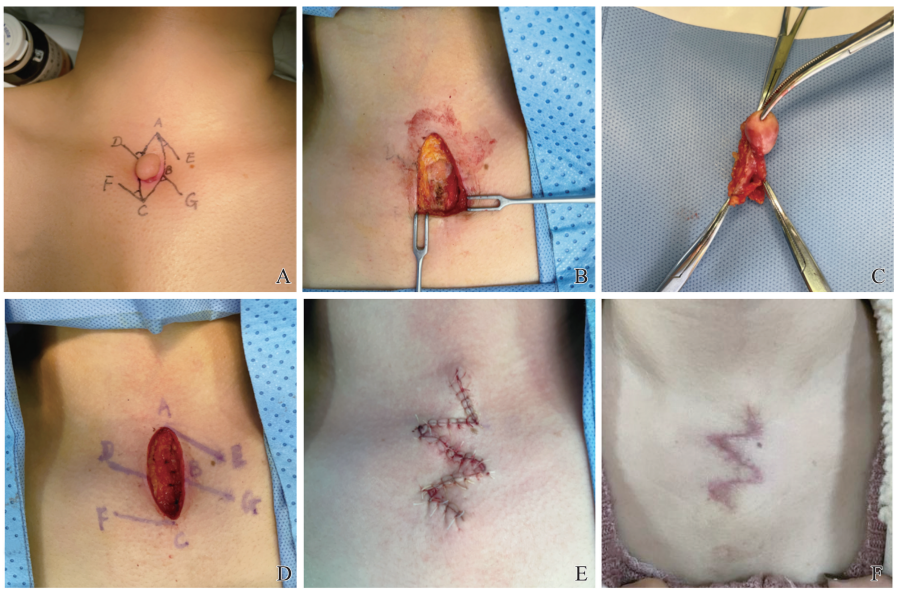

图 2　治疗流程
A:术前皮瓣设计;B:颈前纵行梭形切口;C:切除的肉赘、窦道及皮肤组织;D:术中双 Z 形皮瓣设计; E:“双 Z 成形术”皮瓣修复;F:术后 1 个月切口愈合良好
Figure 2　The treatment process
A: Preoperative flap design; B: Anterior longitudinal spindle-shaped incision; C: Excised fleshy warts, sinuses, and skin tissue; D: Design of the double Z-shaped flap during surgery; E: "Double Z-plasty" flap repair; F: The incision healed well one month after surgery

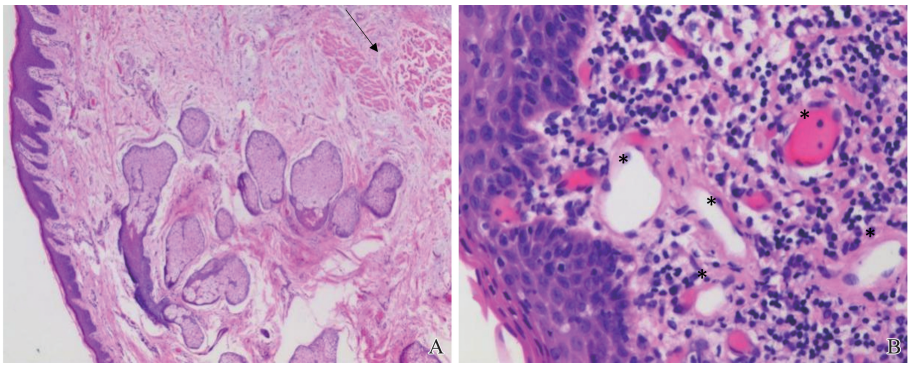

图 3　术后病理图

A：表皮轻度角化亢进，深层可见横纹肌组织（箭头所指）（HE×40）；B：真皮浅层可见管腔形成，管壁内衬鳞状上皮，局灶可见黏液样变性（＊为管腔所在）（HE×100）

Figure 3　The postoperative pathology picture

A: Hematoxylin and eosin staining (×40 magnification) showing mild hyperkeratosis of the epidermis with striated muscle tissue in the deep layer (arrow); B: Hematoxylin and eosin staining (×100 magnification) showing the formation of a lumen (＊) in the superficial layer of the dermis, with the wall lined with squamous epithelium and focal mucinous degeneration

2　讨　论

CMCC 的胚胎起源仍存争议，多数学者认为 CMCC 起源于第二鳃弓在中线的闭合不全；亦有观点认为 CMCC 可能起源于第一/二鳃弓，此种类型常合并下唇、下颌骨、舌及胸骨裂[7-9]。其他发病机制还包括：妊娠早期（第 38 天）头端与心包顶靠近时对颈部的物理挤压[10]；甲状舌管残余[11]；血管的缺血坏死及鳃弓连接部位外胚层组织残留等[12-13]。基因层面研究，CMCC 更倾向于是一类非遗传性偶发畸形[14]，但最近的文献显示，在一组家系的 2 例患者中，发现了相同的可疑突变位点（TYW1B、SSPO）[15]，提示 CMCC 可能存在遗传相关基因。但是由于 CMCC 发病率极低，在现有的报道中仅有一组家系，限制了对此疾病遗传学因素的进一步探索。

CMCC 的典型临床特征是出生即可发现的胸骨上方颈前中线区纵向皮肤裂隙，该裂隙被覆淡红色萎缩皮肤带，上端附着一椭圆形皮赘、下端连接短浅盲窦[8]。常用的影像学手段可被应用于 CMCC 评估。B 超特征为病灶处呈条带状瘢痕纤维突起及无血管的窦道；MRI 表现为颈前中线 T1 低信号 T2 高信号的窦道，不伴有骨性或软骨的改变，部分病例偶见 C6、7 之间椎间盘缺失[9,16-17]。在组织病理学方面，CMCC 多表现为角化过度的复层鳞状上皮，窦道由假复层纤毛柱状上皮和浆液黏液腺组成，真皮层内可有异常存在的横纹肌组织[18]。

CMCC 因其特有的临床体征，多数患儿在新生儿期即可确诊，但临床上仍需与其他外观相似的颈部先天畸形鉴别。第二鳃裂瘘管 80% 以上见于右侧、外瘘口常位于胸锁乳突肌中下段前缘、局部挤压可见透明黏稠分泌物溢出[19-20]；甲状舌管囊瘘的瘘口多位于颈前正中舌骨层面，多为囊肿型继发感染后溃破所致，不存在皮赘及皮肤裂隙改变[21-22]；颈部鳃源性皮肤软骨遗迹好发于左侧，外观多呈棒状皮赘，病灶深面可触及软骨，亦无皮肤裂隙。

CMCC 的治疗目的是彻底根除异常皮肤裂隙，改善颈前外观畸形。出生后 2 年被认为是最佳的手术时机，如合并其他严重畸形时，手术节点可适当前移。CMCC 发病部位位于颈前正中区，为视觉关注的核心美容单元，因此在彻底切除病灶的同时，尚需制定个性化的修复方案。如果患者为颈部皮肤相对松弛的中老年患者，并且病损纵轴较短，可采用单纯的纵向切口，配合仔细的双层缝合，以达到满意的伤口重建[23]。如果患者为儿童（颈部生长最旺盛的年龄为 3~6 岁），并且病损纵轴较长，甚至波及全颈，此时若采用纵向梭形切口和单纯纵向缝合，切口张力过大可继发明显的纵向瘢痕，严重者颈前带状肌挛缩可导致下颌骨后缩等继发畸形[8,24]。尽管已有的研究报告中，最佳修复方法中略存争议，但大多数 CMCC 病例均采用 Z 皮瓣成形术。本病例病灶纵向直径超过 3 cm，并且下缘抵临胸骨上窝，如采用传统的沿颈纹横向梭形切口，术后切口上下张力极大，并且位于骨性结构浅面（胸骨上缘），将不可避免地继发瘢痕。基于最大程度避免并发症，我们采用“双 Z 皮瓣成形术”，此术式的优点为可增加 75% 的皮肤切口轴长[25]，减小张力，避免“单 Z 成形术”皮瓣过大，易扭曲变形的不足。此外，也有学者推荐“W 成形术”，此术式适用于较长的皮肤缺损，虽可避免切口整体延长，但皮瓣翻转度易受限[26]。见图 4。

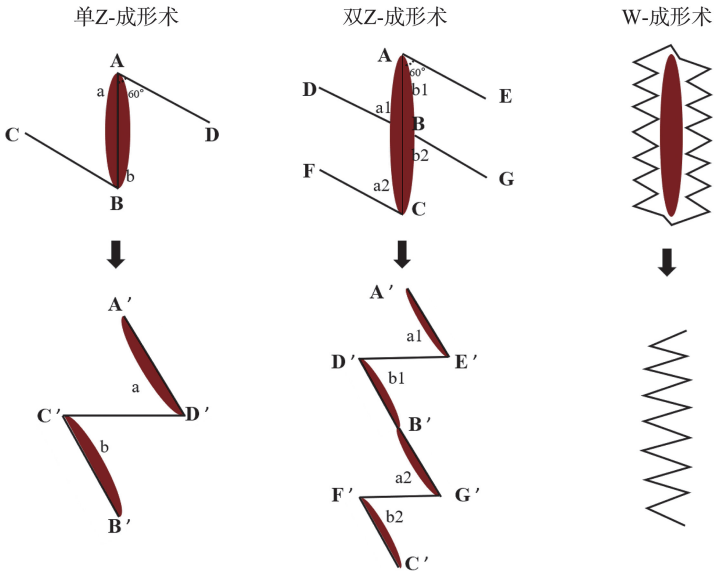

图 4　单 Z-成形术、双 Z 成形术、W-成形术皮瓣修复示意图
Figure 4　Schematic diagram of single Z-plasty, double Z-plasty, and W-plasty for skin flap repair

参考文献：

[1] Eastlack JP, Howard RM, Frieden IJ. Congenital midline cervical cleft: case report and review of the English language literature[J]. Pediatr Dermatol, 2000, 17(2): 118-122. doi:10.1046/j.1525-1470.2000.01727.x

[2] Shaye DA, Nteyumwete H, Kamaliza A, et al. Congenital midline cervical cleft: a rare window to the neck[J]. Facial Plast Surg Aesthetic Med, 2023, 25(2): 184-185. doi:10.1089/fpsam.2022.0101

[3] Puscas L. Midline cervical cleft: review of an uncommon entity[J]. Int J Pediatr, 2015: 209418. doi: 10.1155/2015/209418

[4] 赵利敏, 李晓艳. 先天性颈正中裂 1 例[J]. 中国耳鼻咽喉头颈外科, 2012, 19(4): 224. doi:10.16066/j.1672-7002.2012.04.020
ZHAO Limin, LI Xiaoyan. Congenital median cervical fissure: a case report[J]. Chinese Archives of Otolaryngology-Head and Neck Surgery, 2012, 19(4): 224. doi: 10.16066/j.1672-7002.2012.04.020

[5] 沈为民, 崔杰, 陈建斌, 等, 先天性颈正中裂二例[J]. 中华整形外科杂志, 2009, 25(5): 393. doi: 10.3760/cma.j.issn.1009-4598.2009.05.025

[6] 董玉英, 黄振东, 况光仪. 先天性鳃源性颈正中裂和囊肿 1 例[J]. 中国眼耳鼻喉科杂志, 2002, 2(1): 46. doi:10.3969/j.issn.1671-2420.2002.01.028
DONG Yuying, HUANG Zhendong, KUANG Guangyi. Congenital branchial median cervical fissure and cyst: a case report[J]. Chinese Journal of Ophthalmology and Otolaryngology, 2002, 2(1): 46. doi: 10.3969/j.issn.1671-2420.2002.01.028

[7] Achard S, Leroy X, Fayoux P. Congenital midline cervical cleft: a retrospective case series of 8 children[J]. Int J Pediatr Otorhinolaryngol, 2016, 81: 60-64. doi:10.1016/j.ijporl.2015.12.008

[8] Cudzilo D, Matthews-Brzozowska T, Obloj B. Craniofacial morphology in midline cervical cleft: case report and review of literature[J]. Cleft Palate Craniofacial J, 2017, 54(2): 235-241. doi:10.1597/15-126

[9] Salawu AI, Aremu SK, Olakunle BF, et al. A delayed diagnosis of congenital midline cervical cleft[J]. Clin Case Rep, 2022, 10(3): 05540. doi:10.1002/ccr3.5540

[10] Morton CB. Median cleft of lower lip and mandible, cleft sternum and absence of basihyoid[J]. Arch Surg, 1935, 30(4): 647. doi:10.1001/archsurg.1935.01180100093010

[11] Gross RE, Connerley ML. Thyroglossal cysts and sinuses[J]. N Engl J Med, 1940, 223(16): 616-624. doi:10.1056/nejm194010172231602

[12] Ayache D, Ducroz V, Roger G, et al. Midline cervical cleft[J]. Int J Pediatr Otorhinolaryngol, 1997, 40(2/3): 189-193. doi:10.1016/S0165-5876(97)00028-1

[13] Kara CO, Kara IG. Congenital midline cervical cleft[J]. Otolaryngol Head Neck Surg, 2006, 135(6): 953-954. doi:10.1016/j.otohns.2006.05.024

[14] Jakobsen LP, Pfeiffer P, Andersen M, et al. Genetic studies in congenital anterior midline cervical cleft[J]. Am J Med Genet, 2012, 158A(8): 2021-2026. doi:10.1002/ajmg.a.35466

[15] Masood M.M., Piotr Mieczkowski, Ewa P Malc, et al. Congenital midline cervical cleft: first report and genetic analysis of two related patients[J]. Ann Otol Rhinol Laryngol, 2020, 129(7): 653-656. doi: 10.1177/0003489420906180

[16] Riba M, Bejarano M, Hernández C, et al. Midline cervical cleft: an anatomical finding and a proposal for a new approach[J]. Cleft Palate Craniofacial J, 2020, 57(12): 1422-1427. doi:10.1177/1055665620947957

[17] Goldfisher R, Bawa P, Ibrahim Z, et al. Clinical and

imaging features of a congenital midline cervical cleft in a neonate: a rare anomaly[J]. Case Rep Pediatr, 2015: 439596. doi:10.1155/2015/439596

[18] French WE, Bale GF. Midline cervical cleft of the neck with associated branchial cyst[J]. Am J Surg, 1973, 125(3): 376-381. doi:10.1016/0002-9610(73)90067-6

[19] 黄舒玲, 陈良嗣, 张贝, 等. 双侧先天性第二鳃裂畸形四例[J]. 中华耳鼻咽喉头颈外科杂志, 2016, 51(6): 464-465. doi: 10.3760/cma.j.issn.1673-0860.2016.06.017
HUANG Shuling, CHEN Liangsi, ZHANG Bei, et al. Bilateral congenital second branchial cleft deformity: report of four cases and literature review[J]. Chinese Journal of Otorhinolaryngology Head and Neck Surgery, 2016, 51(6): 464-465. doi: 10.3760/cma.j.issn.1673-0860.2016.06.017

[20] 王莹莹, 张立庆, 周涵, 等. 74 例鳃裂囊肿及瘘管临床分析[J]. 山东大学耳鼻喉眼学报, 2020, 34(4): 111-116. doi: 10.6040/j.issn.1673-3770.0.2019.523
WANG Yingying, ZHANG Liqing, ZHOU Han, et al. Clinical analysis of 74 cases of branchial cleft cysts and fistulas[J]. Journal of Otolaryngology and Ophthalmology of Shandong University, 2020, 34(4): 111-116. doi: 10.6040/j.issn.1673-3770.0.2019.523

[21] 姚丹勉, 陈良嗣, 梁璐, 等. 颈前中线分区清扫治疗儿童复发性甲状舌管囊瘘[J]. 临床耳鼻咽喉头颈外科杂志, 2016, 30(19): 1545-1547. doi:10.13201/j.issn.1001-1781.2016.19.010
YAO Danmian, CHEN Liangsi, LIANG Lu, et al. The selective anterior neck dissection for treating children's recurrent thyroglossal duct cysts[J]. Journal of Clinical Otorhinolaryngology Head and Neck Surgery, 2016, 30(19): 1545-1547. doi:10.13201/j.issn.1001-1781.2016.19.010

[22] 李育广, 刘凤英. 手术治疗甲状舌管囊肿与瘘管 65 例[J]. 山东大学耳鼻喉眼学报, 2008, 22(6): 575-576
LI Yuguang, LIU Fengying. Surgical treatment of 65 cases of thyroglossal duct cyst and fistula[J]. Journal of Otolaryngology and Ophthalmology of Shandong University, 2008, 22(6): 575-576

[23] Annapoorni CV, Varkey S. Congenital midline cervical cleft-A case report on a rare anomaly of the neck[J]. J Indian Assoc Pediatr Surg, 2022, 27: 627-628. doi:10.4103/jiaps.jiaps_206_21

[24] Moreno SD, Christopher P, Kloostra P. Congenital midline cervical cleft (CMCC): z-plasty versus linear cutaneous repair[J]. J Craniofacial Surg, 2022, 34(1): e38-e41. doi:10.1097/scs.0000000000008940

[25] D'Souza JN, Valika T, Maddalozzo J. Surgical management of midline cervical cleft[J]. Int J Pediatr Otorhinolaryngol, 2019, 127: 109657. doi: 10.1016/j.ijporl.2019.109657

[26] Bahakim A, Francois M, Van Den Abbeele T. Congenital midline cervical cleft and W-plasty: our experience[J]. Int J Otolaryngol, 2018: 1-5. doi:10.1155/2018/5081540

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[23] 冯昕, 徐丽娜, 陈文荟, 等. 改良泪前隐窝入路上颌骨囊肿开放术[J]. 中华耳鼻咽喉头颈外科杂志, 2021, 56(8): 863-866. doi:10.3760/cma.j.cn115330-20201118-00875
FENG Xin, XU Lina, CHEN Wenhui, et al. Surgical treatment of maxillary bone cyst though modified endoscopic prelacrimal recess approach[J]. Chinese Journal of Otorhinolaryngology Head and Neck Surgery, 2021, 56(8): 863-866. doi:10.3760/cma.j.cn115330-20201118-00875

[24] 陶谦, 兰天俊. 开窗治疗颌骨囊性病变的临床思考与循证[J]. 口腔疾病防治, 2018, 26(12): 759-765. doi:10.12016/j.issn.2096-1456.2018.12.002
TAO Qian, LAN Tianjun. Clinical thinking and evidence-based for decompression of cystic lesions in the jaw bone[J]. Journal of Dental Prevention & Treatment, 2018, 26(12): 759-765. doi: 10.12016/j.issn.2096-1456.2018.12.002

[25] 范晨旭, 张凯, 陈永锋, 等. CBCT 评价开窗减压术治疗青少年颌骨囊肿的临床疗效[J]. 齐齐哈尔医学院学报, 2019, 40(1): 48-52. doi: 10.3969/j.issn.1002-1256.2019.01.023
FAN Chenxu, ZHANG Kai, CHEN Yongfeng, et al. Evaluation on the clinical effect of decompression in the treatment of juvenile jaw bone cyst by using CBCT[J]. Journal of Qiqihar Medical University, 2019, 40(1): 48-52. doi:10.3969/j.issn.1002-1256.2019.01.023

[26] 孙宁宁, 郭永峰, 周青, 等. 改良引流式开窗术治疗儿童及青少年大型颌骨囊性病变的临床研究[C]//第十四次中国口腔颌面外科学术会议论文汇编. 重庆, 2018: 518.

[27] 郭骏, 黄怡, 费伟, 等. 颌骨囊肿开窗减压术与刮治术临床疗效评价及应用分析[J]. 口腔颌面外科杂志, 2018, 28(4): 219-224. doi:10.3969/j.issn.1005-4979.2018.04.008
GUO Jun, HUANG Yi, FEI Wei, et al. Clinical evaluation and application analysis of fenestration decompression and curettage for jaw cysts[J]. Journal of Oral and Maxillofacial Surgery, 2018, 28(4): 219-224. doi:10.3969/j.issn.1005-4979.2018.04.008

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