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·防治实践·

半侧颜面短小合并心脏及脊柱畸形1例报告及文献回顾

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【摘要】　目的　探讨以半侧颜面短小合并心脏及脊柱畸形为复合表现的先天缺陷患儿的临床特征、诊断思路及多学科治疗策略，为该类复杂病例的临床诊治提供参考。**方法**　回顾性分析1例9岁半侧颜面短小畸形(hemifacial microsomia，HFM)合并法洛四联症术后及脊柱侧弯患儿的临床资料。通过专科检查、影像学检查、骨龄测定及智力评估等明确诊断，患儿表现为右侧HFM(伴3个副耳、面横裂、上唇正中微小型唇裂、下颌骨及面部软组织发育不良、右侧腮腺及喙突缺如)、眶距增加、牙列紊乱、1颗侧切牙先天缺失、乳恒牙猖獗龋；4年前因法洛四联症合并卵圆孔未闭行开胸心脏手术；并存在脊柱侧弯及全身发育迟缓(骨龄约7岁)。结合文献对该类型病例的诊疗进行回顾性分析。**结果**　采用多学科联合诊疗(multi-disciplinary treatment，MDT)模式，对该患儿先行口腔龋病治疗，后在全身麻醉下行右侧副耳切除、面横裂及上唇微小型唇裂修复术。经6个月随访，患儿面部外形显著改善，口腔功能恢复良好。文献回顾结果表明半侧颜面短小畸形是一种以半侧面部多种组织结构发育不良为特点的先天性疾病，病因可能与妊娠早期第一、二鳃弓血供障碍有关，常累及颅面骨、耳及软组织，导致呼吸、进食、语言、听力等功能障碍及心理问题，严重者影响生存质量。合并心脏与脊柱畸形者较为罕见，需根据临床评估与手术指征制定个体化序列治疗方案，涵盖心脏外科手术、脊柱矫形、早期软硬组织重建(牵张成骨、面裂修复、副耳切除等)、生长发育期正畸与牙列管理以及成年后的轮廓修整。**结论**　HFM可合并心脏与脊柱畸形，临床表现复杂；早期诊断、MDT协作及序列治疗是改善预后与生活质量的关键。

【关键词】　半侧颜面短小畸形；　第一二鳃弓综合征；　心脏病；　法洛四联症；　脊柱侧弯；　发育缺陷；　手术修复；　多学科联合诊疗

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Hemifacial microsomia with cardiac and vertebral anomalies: a case report and literature review

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【Abstract】　Objective　To investigate the clinical characteristics, diagnostic approach, and multidisciplinary treat-

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ment strategy for a rare case of congenital defect presenting as a complex of hemifacial microsomia with cardiac and spinal deformities, in order to provide a reference for the clinical management of such cases. **Methods** The clinical data of a 9-year-old patient with hemifacial microsomia (HFM) complicated by post-operative Tetralogy of Fallot and scoliosis were retrospectively analyzed. A definitive diagnosis was established through specialized examinations, imaging studies, bone age assessment, and intellectual evaluation. The patient presented with right-sided HFM (with 3 accessory auricles, a transverse facial cleft, a microform median cleft of the upper lip, hypoplasia of the mandible and facial soft tissues, and agenesis of the right parotid gland and coronoid process), increased orbital distance, dental malalignment, congenital absence of one lateral incisor, and rampant caries in both primary and permanent dentition. The patient had undergone open-heart surgery for Tetralogy of Fallot with a patent foramen ovale four years prior and also presented with scoliosis and systemic developmental delay (bone age approximately 7 years). A retrospective analysis of the diagnosis and treatment of this type of case was conducted in conjunction with a literature review. **Results** A multi-disciplinary treatment (MDT) model was adopted. The patient first received treatment for dental caries, followed by excision of the right accessory auricles, repair of the transverse facial cleft, and correction of the microform upper lip cleft under general anesthesia. A 6-month follow-up showed significant improvement in facial appearance and good recovery of oral function. The literature review indicated that hemifacial microsomia is a congenital disease characterized by the hypoplasia of multiple tissue structures on one side of the face. Its etiology may be related to impaired blood supply to the first and second branchial arches during early pregnancy. It often affects the craniofacial bones, ears, and soft tissues, leading to functional impairments in respiration, feeding, speech, and hearing, as well as psychological issues, severely impacting the quality of life in serious cases. The combination with cardiac and spinal deformities is relatively rare and requires individualized sequential treatment plans based on clinical evaluation and surgical indications. This typically includes cardiac surgical correction, spinal orthopedics, early soft and hard tissue reconstruction (e.g., distraction osteogenesis, facial cleft repair, and accessory auricle excision), orthodontic and dental management during the growth period, and final facial contouring in adulthood. **Conclusion** HFM can be associated with cardiac and spinal deformities, presenting with complex clinical manifestations. Early diagnosis, MDT collaboration, and sequential treatment plans are key to improving patients' prognosis and quality of life.

【Key words】 hemifacial microsomia; first and second branchial arch syndrome; cardiac disease; Tetralogy of Fallot; scoliosis; developmental defects; surgical repair; multi-disciplinary treatment

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半侧颜面短小畸形(hemifacial microsomia,HFM)是一种以单侧面部多种组织结构发育不良为特征的先天性疾病[1],主要临床表现为颅面骨、耳廓及面部软组织发育不良。其发病率为1.8~2.9/10 000,是发病率仅次于唇腭裂的常见颅面部先天性畸形[2-3]。HFM发病机制与第一、二鳃弓的发育异常相关[4]。HFM不仅造成严重的面部畸形,还可伴随呼吸、进食、语言、听力等功能障碍及心理障碍[5-7]。畸形严重时可显著降低患者的生存质量,并为其家庭带来沉重的心理与经济负担,因此通常需多学科协作进行系统性、序列化的综合治疗[8-10]。文献检索发现,目前尚未见同时罹患半侧颜面短小畸形、脊柱畸形与心脏畸形的综合征型病例报道。现有文献多集中于与第一、二鳃弓发育异常相关的综合征及其并发表现[5-6],如Goldenhar综合征、耳髁综合征、Treacher collins综合征、Pierre Robin序列征等[10]。本文报道的患者存在多种口腔颌面部异常表现,包括右侧副耳、面横裂、上唇微小型唇裂、眶距增大、牙列不齐、乳恒牙猖獗龋、1颗侧切牙缺失、右侧腮腺缺如、右侧喙突缺如、右侧颜面部短小萎缩畸形等,符合第一二鳃弓综合征的临床特征。然而,其合并先天性心脏病、脊柱侧弯及全身发育迟缓的复杂表型,尚未见文献报道。因此,本文回顾性分析1例临床表现为半侧颜面短小合并心脏及脊柱畸形复合症状患儿的诊疗过程,以期为该类罕见复杂病例的临床

诊断与综合管理提供借鉴。

1　病例资料

1.1　一般资料

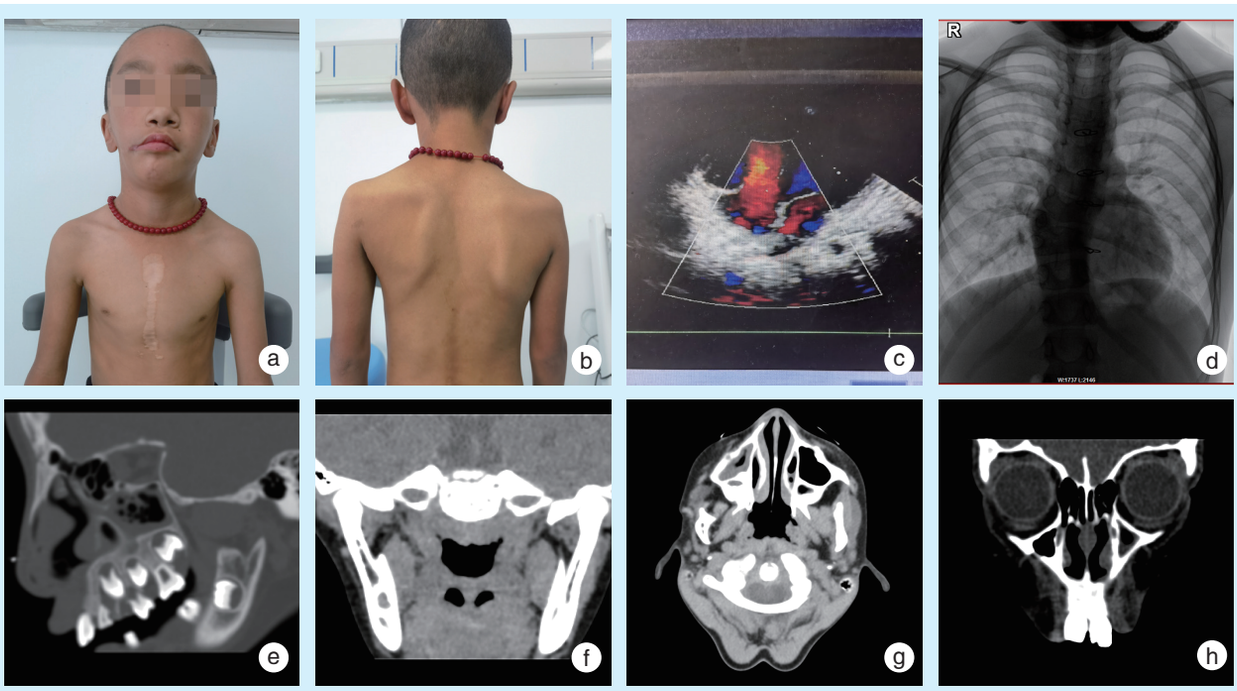
患者，男性，9岁，因“先天性右侧耳前赘生物及面部畸形”至阿坝藏族羌族自治州人民医院口腔科就诊，患者4年前因法洛四联症合并卵圆孔未闭，接受胸骨正中切口行法洛四联症全部修补术+卵圆孔缝闭术+动脉导管结扎术，术后定期随访6个月，复查心电图未见异常，恢复良好。家族史：父母与弟弟均体健，无类似系统性疾病及遗传病史。

1.2　临床检查

1.2.1　体格检查　胸前见手术瘢痕，脊柱侧弯，斜肩姿势，无法完全直立，身高低于同龄人，整体发育不良。

1.2.2　专科检查　双侧颜面不对称，右侧颜面部发育不良，伴3个右侧耳前赘生物；右侧腮腺及喙突未能触及，软组织萎缩；眶距增宽，外眦外展，右侧鼻翼上抬、鼻小柱右偏；右侧面横裂伴口角歪斜，皮肤可见线性痕迹，裂隙宽度可从口角直至颊前1/3，和副耳牵连；右上红唇凹陷，唇峰较对侧短小。口内见乳恒牙猖獗龋，牙列不齐，右侧恒牙萌出迟缓。

1.2.3　影像学检查　口腔全景片及锥形束CT（cone beam computed tomography，CBCT）显示双侧下颌骨不对称，右侧腮腺缺如、喙突缺如，22牙缺失，双侧恒牙萌出不对称，多颗乳恒牙龋坏，处于替牙期，气道通畅。头颅、颌面部、胸部螺旋CT（平扫+重建）：右侧耳前3个副耳、面横裂、上唇正中微小型唇裂、眶距增宽；右侧腮腺缺如，右侧喙突缺如，右侧颜面部短小畸形；正中开胸手术后瘢痕改变，未见明显异常。颈椎正位片：脊柱侧弯（图1）。



a: anterior views of the thorax and spine, showing a midline sternotomy scar from prior cardiac surgery and significant scoliosis; b: posterior views of the thorax and spine, showing a midline sternotomy scar from prior cardiac surgery and significant scoliosis; c: echocardiogram showing mild tricuspid and mitral regurgitation with intact septa; d: radiograph confirming scoliosis and post-cardiac surgery status; e: CBCT (sagittal view, bone window) demonstrates absence of the right coronoid process and hypoplasia of the right mandible; f: CBCT (coronal view, soft tissue window) reveals absence of the right parotid gland. The base of an accessory auricle is situated in the parotid region, and the right mandibular ramus is shortened compared to the left; g: CBCT (axial view, soft tissue window) confirms absence of the right parotid gland, with the accessory auricle base located in the parotid region. The right side shows reduced soft tissue fullness relative to the left; h: CBCT (coronal view, soft tissue window) shows an interorbital bony distance of 30 mm, exceeding the normal pediatric range (20 – 25 mm), consistent with orbital hypertelorism

Figure 1　Hemifacial microsomia in a patient with cardiac and vertebral anomalies

图1　半侧颜面短小患者合并心脏及脊柱畸形

1.2.4　其他辅助检查　心电图：窦性心律，心脏瓣膜术后改变。骨龄检查：发育延迟，相当于7岁水平。韦氏儿童智力量表（Wechsler intelligence scale for children-revised，WISC-R）测试：89分（智力评分正常）。

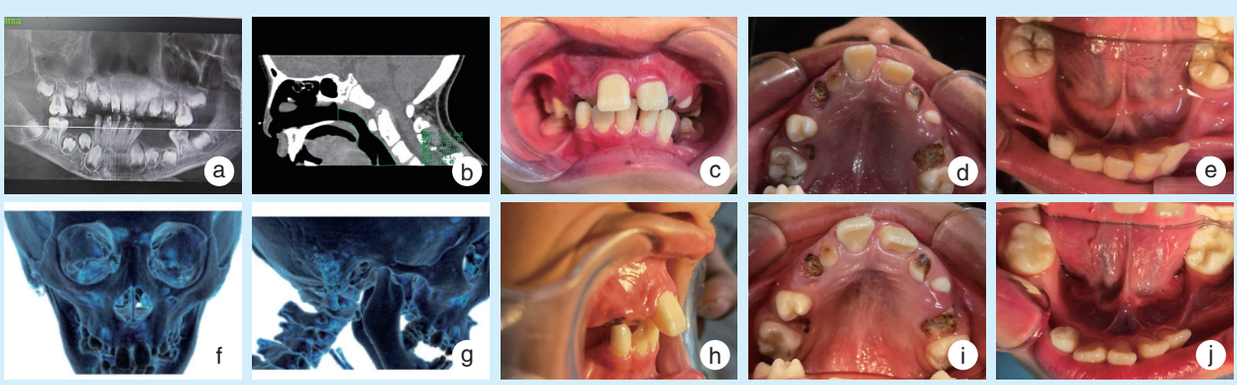
1.3　诊断

根据患者的临床表现、影像学检查及家族史，该例患儿诊断为“半侧颜面短小畸形合并心脏与脊柱畸形”。

1.4　治疗

多学科联合诊疗（multi-disciplinary treatment，MDT）模式为该患儿制定个性化序列治疗方案。治疗分为以下阶段实施：首先在口腔门诊进行全口恒牙龋坏充填修复及窝沟封闭，并进行系统的口腔卫生指导；随后于全身麻醉下行右侧副耳切除术、面横裂修复术及微小型唇裂整复术，以改善颌面部外形，术后第3天患者顺利出院。见图2、3。

出院后对患儿进行定期随访与口腔健康管理。同时建议家属后续对脊柱侧弯进行专科矫治，并计划在未来阶段进行右侧腮腺区软组织重建及正畸-正颌联合治疗。经3个月及6个月随访，患儿颌面部外形显著改善，恒牙修复效果良好，口腔功能和外貌均得到有效改善。此外，患儿的全身状况也有改善，家属对治疗效果满意。



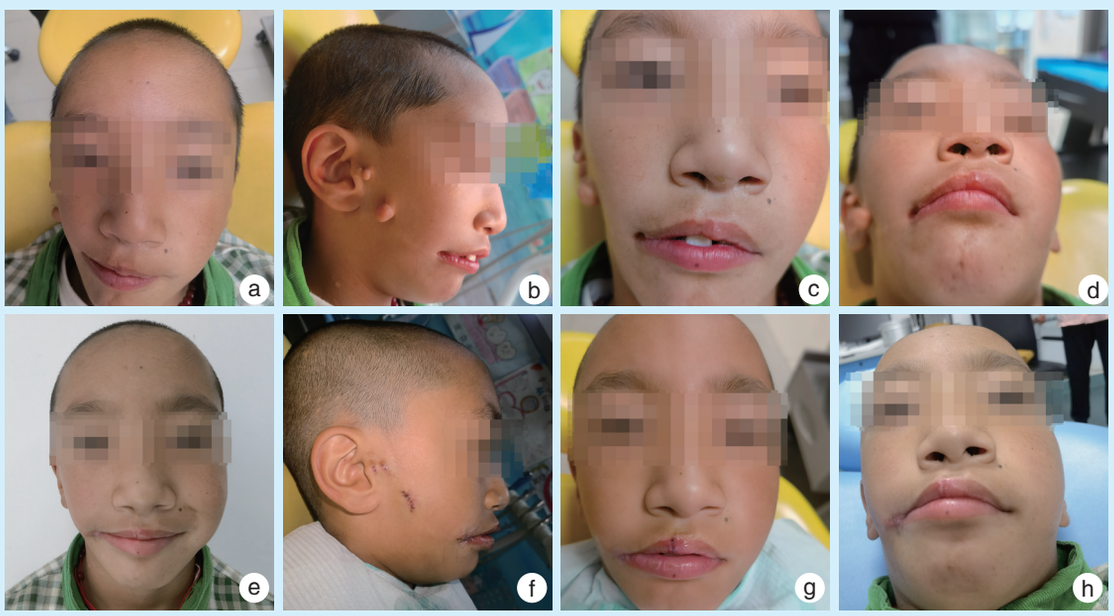
a: oral panoramic tomography shows that tooth number 22 is missing, with the permanent teeth on both sides erupting asymmetrically. Multiple primary and permanent teeth have caries, and the patient is in the mixed dentition stage. b: CBCT (cone beam computed tomography) airway reconstruction analysis indicates that the airway is unobstructed, with no stenosis. c: intraoral bite reveals rampant caries in primary and permanent teeth, malocclusion, deep overbite, and delayed eruption of permanent teeth on the right side. d: intraoral upper jaw dentition shows multiple deciduous tooth root remnants and missing teeth, with caries in the pits and fissures of teeth 16 and 26. e: intraoral lower jaw dentition shows multiple deciduous teeth missing, with caries in the pits and fissures of teeth 36 and 46. f: CBCT three-dimensional reconstruction in the coronal view shows underdevelopment of the right zygomatic region and right mandible, with asymmetry between the left and right sides. g: CBCT three-dimensional reconstruction shows the absence of the right coracoid process, and the mandible is shorter and smaller than the left side. h: three days after postoperative intraoral lateral decubitus shows a deep overbite. i: three days after postoperative fissure sealant application for permanent maxillary teeth in the oral cavity. j: three days after postoperative fissure sealant application for permanent teeth in the lower jaw inside the mouth

Figure 2　Pre- and post-treatment intraoral photographs and CBCT imaging of hemifacial microsomia in a patient with cardiac and vertebral anomalies

图2　半侧颜面短小合并心脏及脊柱畸形患儿治疗前后口内像及CBCT影像

2　讨　论

HFM又称第一、二鳃弓综合征，是指由第一、二鳃弓及其间的第一鳃裂和颞骨始基先天性发育异常引起的一系列畸形[2-3, 10]。本病男女发病率之比为3∶2[4, 11]，右侧与左侧发病率约3∶2[4, 11]，约有10%患者同时伴发唇腭裂[12-13]。其病因和发病机制目前尚未明确，由于HFM表型及遗传高度异质性，染色体畸变类型、基因组亚微观改变及致病基因等检出率低，且结果重复性差[2-3, 11]。HFM的临床表现多样，主要包括：①外耳畸形[7-8, 14]，常伴发副耳、耳前瘘以及外耳道、中耳、内耳的结构异常，耳廓形态异常、缺失、位置改变，外耳道闭锁，中耳骨链异常，导致不同程度的传导性听力损失；②半侧颌骨短小畸形[5, 15-18]，表现为同侧或单侧下颌骨发育不足[19]，上颌垂直向发育不足，甚至颞下颌关节消失而形成假关节，咬合关系紊乱，咀嚼肌和腭肌发育不全，引起咀嚼功能障碍，严重下颌后缩可引起呼吸障碍，因气道阻塞需要侧卧、俯卧等，可



a-d: pre-operative views showing bilateral facial asymmetry with right hemifacial hypoplasia and soft tissue atrophy. Note the three right preauricular tags, hypertelorism, lateral displacement of the outer canthus, elevated right nasal ala, and deviated columella. A right transverse facial cleft extends from the oral commissure, which is associated with a linear skin groove and a deviated corner of the mouth. The right upper vermilion border is depressed with a shortened philtral column. e-h: three days after complete excision of preauricular tags and repair of the transverse facial cleft and microform cleft lip, showing significant improvement in bilateral facial asymmetry deformities, correction of the crooked right mouth corner, disappearance of right accessory ear, and basic symmetry of the upper lip vermilion and peak

Figure 3　Pre- and post-operative facial photographs of hemifacial microsomia in a patient with cardiac and vertebral anomalies

图3　半侧颜面短小合并心脏及脊柱畸形患儿颜面部术前术后颌面部外观对比

能需要气管切开开放气道、牵引舌体防止后坠、下颌骨牵张成骨或颏前徙延长颌骨长度[10]；③口裂畸形[12, 20-21]，如大口畸形、面横裂和面斜裂，可出现唇腭裂、半侧舌体发育不全；④软组织缺损[2-3]，主要是由患侧肌肉、脂肪发育不全造成的，也见腮腺缺如或异位，颊部瘘管；⑤牙萌出异常[4]，包括先天牙缺失（不包括第三磨牙），及患侧牙齿萌出延迟；⑥眼部异常[7]，如眼睑下垂、睑裂过小，眼球运动障碍；⑦面神经功能障碍[2-3]，可累及颞支、颧支、上下颊支、下颌缘支、颈支，导致相应区域的面瘫；⑧中枢神经系统异常[2-3]，表现为智力发育迟缓或认知障碍；⑨其他系统畸形，如心脏、肾脏、肋骨或脊柱发育异常等。上述症状并非全部出现于同一患者，其中以外耳畸形和半侧颌骨短小畸形最为常见。本例患者符合第一二鳃弓综合征的表现，但同时出现喙突缺如、腮腺缺如、心脏及脊柱畸形、骨龄延迟及全身发育和智力缺陷，拓展了该病的表型谱，也是对第一二鳃弓综合征分型的进一步补充与完善[22-23]。

对于患者出现的软硬组织畸形，本文重点讨论腮腺缺如这一表现。先天性涎腺缺失或发育不全是一种原因不明的罕见先天性疾病，少数患者有家族史，提示可能与遗传缺陷有关[24-25]。该病发生于任何大小唾液腺，单双侧均可出现，其中腮腺相对常见。患者还可能伴随头颈部其他异常，如副耳、鳃弓综合征、泪器异常、先天性牙缺失、过小牙及骨畸形等，也可见于外胚叶发育不全的个体[12]。其临床特点包括出生后唾液分泌减少，口干或口腔黏膜干燥，检查时无法定位相应唾液腺导管开口，常伴牙猖獗龋、白念珠菌感染、咽喉炎及黏膜炎等症状[11, 18]。

该病常与肢端、皮肤、指（趾）甲、泪腺、牙齿综合征（acro-dermo-ungual-lacrimal-tooth syndrome，ADULT综合征）相关，这是一种典型的外胚层发育异常综合征，与p63基因的错义突变有关；也和泪-耳-牙-指综合征（lacrimo-auriculo-dento-digital syndrome，LADD综合征）相关，一种少见的常染色体显性遗传疾病，表现为泪囊、耳廓、涎腺和骨骼系统畸形，也有肾脏异常、食管裂孔、面部畸形及会厌发育不全等症状[26]。本例患者部分症状和涎腺缺失罕见病相同，但未能完全涵盖所有特征。综合分析认为，患者的口干、猖獗龋、面部软组织

畸形等症状可能与腮腺缺如导致涎腺功能异常有较大关系。鉴于患儿年龄较小，实施腮腺解剖、功能及美学重建存在较大困难，目前通过窝沟封闭及龋齿充填修复进行预防干预，并指导患者加强口腔卫生、保持口腔湿润等口腔习惯及健康宣教。本病例也为临床检查、诊断及治疗提供新的思路。

通过对本病例患者的系统分析，并结合相关文献及案例报道进行鉴别诊断，笔者通过详细询问病史、实施全身检查，充分掌握了其临床表现及疾病进展，进而制定了合理的治疗方案。因患者先天性心脏病已手术痊愈，本次治疗主要针对口腔及颜面部病变进行对症处理。已向患者家属详细告知病情的复杂性，说明需要结合骨科、颌面外科、整形外科、正畸科、正颌外科及牙体牙髓科等多学科协作，实施长期、有序的综合治疗。建议尽早矫正脊柱侧弯畸形，二期评估后开展右侧腮腺软组织重建修复；后续通过正畸-正颌联合治疗及牵张成骨等技术，矫正颌骨发育不良及牙列畸形；最终对面部瘢痕及不对称畸形进行美容修复，完成序列治疗[27-30]，以改善半面短小畸形及牙列不齐。

文献报道此类患者可合并气道狭窄甚至呼吸睡眠暂停低通气综合征（obstructive sleep apnea，OSA）[31]。本例患者经气道测量及临床病史回顾，未见气道狭窄表现，故未联合耳鼻喉科进行早期气道干预或扩宽治疗。研究表明60%上、下颌骨发育异常为常染色体显性遗传，耳廓及颜面部软组织发育畸形多无遗传史[32]。该病例未对患者及其直系亲属进行基因检测及深入的家族史调查，因此未能明确是否存在遗传学风险。

综上所述，本文报道了1例半侧颜面短小畸形合并心脏及脊柱异常的患者，详细呈现其专科与全身检查、临床表现、诊断及初步治疗过程。通过实施个性化治疗方案，患者颌面部外形显著改善。该疾病较为罕见，早期发现、及时干预与针对性治疗具有重要意义。

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