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Clinical and Diagnostic Aspects of Submucous Cleft Palate in the Practice of the Otorhinolaryngologist and Maxillofacial Surgeon

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ABSTRACT

BACKGROUND: Submucous cleft palate is an uncommon type of isolated clefts. Its diagnosis is not challenging: a triangular pit due to bone loss along the midline of the hard palate; a translucent mucosal duplication region in the midline soft palate, causing its muscle impairment, nasalization, and a bifid uvula. In case of the compensated submucous cleft palate and unclear clinical signs, diagnosis is challenging.

AIM: To determine clinical signs (markers) of X-ray computed tomography and magnetic resonance imaging for the diagnosis of submucous cleft palate.

METHODS: A retrospective analysis of 21 medical records of patients with submucous cleft palate was conducted in 2019–2024. All patients underwent conservative and surgical treatment under the compulsory health insurance plan. All patients underwent X-ray computed tomography or magnetic resonance imaging.

RESULTS: Magnetic resonance imaging showed a linear hypointense structure along the midline due to the intermittent levator muscles of the soft palate. X-ray computed tomography identified three typical markers of submucous cleft palate, including a triangular palate defect on a 3D reconstructed image of the skull; a palate defect in the frontal view and a shortened vomer; anterior displacement of the posterior nasal spine and a large nasopharyngeal space in the sagittal view. Patients seek medical help for upper airways infections from an otolaryngologist much earlier. Our study showed significant differences in the age of diagnosis of the submucous cleft palate by otorhinolaryngologists and other medical professionals ($p = 0.015$).

CONCLUSION: Otorhinolaryngologist can detect manifestations and effects of submucous cleft palate and suspect the defect much earlier than other medical professionals. A promising path in identifying submucous cleft palate is to use radiologic imaging methods in routine practice. Timely detection of the submucous cleft palate will allow earlier rehabilitation to improve the quality of life and speech.

Keywords: submucous cleft palate; X-ray computed tomography; magnetic resonance imaging.

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Оригинальное исследование

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Клинические и диагностические аспекты подслизистой расщелины нёба в практике оториноларинголога и челюстно-лицевого хирурга

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АННОТАЦИЯ

Обоснование. Подслизистая расщелина нёба — нечастая форма изолированных расщелин. Диагностика ее не представляет трудности: треугольная ямка из-за костного дефекта по средней линии твердого нёба, полупрозрачная зона дупликатуры слизистой оболочки в срединной части мягкого нёба, обеспечивающая недостаточность его мышц и назализацию голоса, расщепленный язычок. При компенсированной форме подслизистой (скрытой) расщелины нёба и неявно выраженных клинических признаках возникают диагностические трудности.

Цель — установить клинические признаки-маркеры рентгеновской компьютерной томографии и магнитно-резонансной томографии для диагностики подслизистой расщелины нёба.

Методы. Проведен ретроспективный анализ 21 истории болезни пациентов с подслизистой расщелиной нёба в период с 2019 по 2024 г. Все пациенты проходили консервативное и хирургическое лечение в рамках обязательного медицинского страхования. Всем пациентам проводили рентгеновскую компьютерную или магнитно-резонансную томографию.

Результаты. По данным магнитно-резонансной томографии можно наблюдать по средней линии линейную гипоинтенсивную структуру вследствие прерывистого положения мышц-леваторов мягкого нёба. По данным рентгеновской компьютерной томографии выделены три характерных признака подслизистой расщелины нёба: треугольный дефект нёба на 3D-реконструкции черепа, дефект нёба во фронтальной плоскости и укорочение сошника, смещение задней носовой ости кпереди и увеличение пространства носоглотки в сагиттальной проекции. Пациенты гораздо раньше начинают обращаться за помощью в лечении инфекции верхних дыхательных путей к оториноларингологу. Согласно полученным данным настоящего исследования, при оценке возраста обнаружения подслизистой расщелины нёба ЛОР-врачом и другими специалистами были выявлены существенные различия (p=0,015).

Заключение. Оториноларингологи гораздо раньше других специалистов могут столкнуться с проявлениями и следствием подслизистой расщелины нёба и заподозрить наличие порока. Перспективное направление в выявлении этой патологии — использование лучевых методов диагностики в постоянной практике. Своевременное обнаружение подслизистой расщелины нёба позволит раньше начать реабилитационные мероприятия по улучшению качества жизни и речи.

Ключевые слова: подслизистая расщелина нёба; рентгеновская компьютерная томография; магнитно-резонансная томография.

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ЭКО • ВЕКТОР

BACKGROUND

Various types of congenital cleft palate are the most frequent facial skeleton defects. These clefts require correction at an early age, factoring in the patient's individual characteristics and health status. The treatment of and rehabilitation from this condition involve not only maxillofacial surgeons but also an otorhinolaryngologist, audiologist, pediatrician, geneticist, speech-language pathologist, orthodontist, dentist, psychologist, and other related professionals [1, 2].

A submucous cleft palate (SMCP) is a rare form of isolated clefts characterized by intact palatal mucosa despite underlying muscle impairment. In his monograph, Mamedov [3] notes that the first documented cases of this pathology were published in 1825 by Roux; Demarquay provided an anatomical description of this defect in 1846; Kelly introduced the term "submucous cleft palate" in 1910, which still remains in use in English-language medical literature today.

In case of SMCP involving the soft palate, only the soft palate muscles are split along the midline, whereas the bony structures of the palate and mucosa remain intact. In cases of SMPC involving both the hard and soft palate, a midline groove (which increases during speech and contraction of cleft soft palate muscles) and a bifid uvula are observed; the palate may be shortened. The mucosa constitutes a duplication of nasal and oral mucosas, with its thinning and translucency clearly visible during fiberoptic nasopharyngoscopy. Palate palpation may reveal non-union of the maxillary palatine processes along the midline. Clinically, patients exhibit varying degrees of hypernasal voice [4].

Stal and Hicks [5] classified three types of SMCP based on the position of levator muscles: Type A (most muscle fibers attach to the hard palate); Type B (a moderate portion of muscles attaches to the hard palate); Type C (a minimum portion of muscles attaches to the hard palate).

SMCP diagnosis is not challenging, where three major symptoms are in existence: a triangular pit due to bone loss along the midline of the hard palate; a translucent region in the midline soft palate, or *zona pellucida*; associated soft palate muscle impairment, causing nasalization and a bifid uvula.

Extra symptoms in favor of SMCP diagnosis may include: hearing impairment and presence of secretory otitis media, recurrent otitis media with effusion, as the soft palate muscles also regulate the auditory tube function, maintaining its protective, drainage, and ventilatory roles [3, 6]. Multiple researchers described the anatomical pre-requisites for auditory tube dysfunction, with the cleft itself being attributed to risk factors for developing secretory otitis media, chronic otitis media with cholesteatoma, and hearing impairment [7].

Upon reviewing foreign literature, the authors found no clinical reviews or articles specifically addressing the diagnosis and treatment of patients with SMCP. They found small studies with limited sample sizes, focusing either on surgical techniques for patients with SMCP or on documenting the observation of a rare syndrome associated with SMCP. Russian literature contains only isolated cases outlined in thesis papers or articles devoted to cleft palate. No clinical reviews with large-scale observations over patients with SMCP were identified either: the largest reported patient group consisted of just 7 people [8].

Prior to the widespread adoption of instrumental diagnostic methods, SMCP diagnosis relied upon the identification of traditional physical examination symptoms coupled with evidence of velopharyngeal insufficiency [9, 10]. However, in case of the compensated SMCP, there is no pronounced nasalization, which renders the diagnosis challenging.

According to Finkelstein et al. [11], nasal endoscopic examination provides valuable insights into velopharyngeal valve physiology and pathology. However, performing endoscopy in young children is difficult. In their study of 40 patients, the researchers demonstrated how nasal endoscopy contributes extra diagnostic information about palatal musculature abnormalities.

Researchers are split over assessment methods, criteria to be followed in diagnosing SMCP, and the appropriate age for SMCP repair. Thus, the 20-year retrospective study done by Ten Dam et al. [12] covered 28 non-syndromic patients diagnosed with SMCP. The average age at diagnosis was 3.9 years, and all patients had one or several symptoms: hypernasal voice (65%), articulation difficulties (46%), conductive hearing impairment (39%), and/or swallowing issues (32%). A bifid uvula was discovered in 92% cases. Postoperatively, rhinolalia and swallowing issues were resolved, hearing improved (though without complete normalization). Articulation difficulties remained.

The research done by Kuehn et al. [13] demonstrated the potential of magnetic resonance imaging (MRI) as the only method of visualizing the *m. levator veli palatini*, being in charge of palatal elevation. MRI-based studies remain very limited, but researchers successfully assessed different positions of the levator muscle in normal versus abnormal anatomical configurations. Researchers also employed MRI to compare the muscle elevating the palatine velum in post-palatoplasty patients with congenital cleft palate and normal participants [14]. Literature primarily outlines isolated clinical cases of using MRI for SMCP diagnosis in the form of intermittent levator muscles and subsequent surgical intervention demonstrating positive speech outcomes [15].

In their study of 28 patients with auditory tube dysfunction, Bae et al. [16] for the first time identified the bony notch defect in the hard palate and the associated

vomer defect in 4 patients through the use of X-ray computed tomography (CT). On top of that, the hard palate was shorter in patients with the auditory tube dysfunction versus the control group ($p = 0.016$).

The experience of Andreeva et al. [17] of using CT for SMCP diagnosis was published. However, the CT method is effective for patients with clefts of the hard palate. Three CT markers enable accurate SMCP identification and explain anatomical particularities of pathology of intranasal structures, nasopharynx, and middle ear. CT may also detect patulous auditory tubes and middle ear abnormalities to manage surgical planning.

McWilliams [18] performed a retrospective analysis of 130 patients with SMCP, with 56% underwent surgical correction, whereas 44% required no intervention due to the absence of clinically significant symptoms.

Cases of fistula occurring in the SMCP region and causing nasal regurgitation in patients of various age groups were outlined. Such fistulae were successfully repaired using isolated cleft palate reconstruction principles, with no fistula recurrence reported [19, 20].

According to a retrospective study of 72 patients with SMCP, surgical repair was performed in 46 cases at late stages, 49.74 months after establishing the anatomical reason of speech disorders, on average. The authors believe that early repair reduces the likelihood of further intervention, while improving the expectations of positive postoperative speech outcomes [21].

Literature reported the case of delayed SMCP diagnosis at the age of 25, even with hypernasal speech and nasal food regurgitation. The patient examination did not reveal any audiologic disorders, syndromes and malfunctions, deviations in the form of physical developmental / speech delays. It is worth mentioning delayed diagnosis and, accordingly, late-stage rehabilitation of the patient, particularly given that the specified clinical case was published in a US-based journal in 2020 [22].

SMCP diagnosis requires that otorhinolaryngologists perform adenoidectomy to account for this malfunction. In reliance upon the 1993–2003 formal analysis of 20 patients with SMCP, who underwent transnasal partial endoscopic adenoidectomy, Stern et al. [23] demonstrated that this technique is both safe and highly effective.

Nevertheless, diagnosing SMCP is challenging even with known clinical symptoms owing to the pathology's rarity, weakly noticeable visual indicators, insufficient clinician awareness, and infrequent use of radiologic imaging methods for SMCP diagnosis. Otorhinolaryngologists must be aware of SMCP; incorporate recommendations and diagnostic algorithms into routine practice, placing particular emphasis on chronic inflammatory processes in the middle ear and nasopharynx; timely diagnose SMCP to prevent the worsening of rhinolalia during adenoidectomy; perform simultaneous middle ear surgeries to avoid severe hearing impairment.

The study is aimed at determining clinical signs (markers) of CT of paranasal sinuses and MRI of the maxillofacial region for the diagnosis of SMCP.

METHODS

A retrospective analysis in respect of 21 medical records of patients with SMCP was conducted in 2019–2024. All patients underwent conservative and surgical treatment under the compulsory health insurance plan at the Department of Otorhinolaryngology and/or Maxillofacial Surgery of State Autonomous Healthcare Institution Children's Republican Clinical Hospital under the Ministry of Health of the Republic of Tatarstan. SMCP diagnosis was the criterion for inclusion in the analysis. All patients completed CT or MRI, standard clinical laboratory tests, and instrumental examinations. Patients underwent otorhinolaryngologic follow-up for 1–5 years.

Statistical analysis was performed using the StatTech v. 4.2.6 software (Russia), with statistical significance set at $p < 0.05$.

RESULTS

The median (*Me*) age at SMCP diagnosis was 7.62 ± 3.85 years; boys prevailed (66.7%), and there were 7 girls (33.3%). A submucous cleft involving both the hard and soft palate was diagnosed in 20 patients, of which 19 patients with midline clefts, one with a right-sided cleft, and one with a cleft of the soft palate only. Fiberoptic rhinopharyngoscopy revealed a nasopharyngeal enlargement due to posterior nasal cavity expansion driven by vomer shortening and a clear visualization of contralateral posterior nasal structures ($n = 19$). A distinct midline soft palate pit is observed in 12 patients.

Otorhinolaryngologists diagnosed SMCP in 15 cases (71.4%) for the first time. As per the data obtained, significant differences in the age of SMCP diagnosis by otorhinolaryngologists and other medical professionals were revealed ($p = 0.015$) (the method used is the Student's *t*-test). Whereas patients with hypernasal speech typically seek medical help from speech therapists or maxillofacial surgeons earlier, otorhinolaryngologists are in a position to detect SMCP manifestations much sooner and, knowing about this condition, must determine timely rehabilitation tactics.

To diagnose a paranasal sinus, middle ear pathology, and SMCP, 19 patients underwent CT imaging. In reliance upon the CT data, the authors of this article identified the following three typical CT markers of SMCP.

1. A triangular defect of varying degrees is clearly seen on a 3D reconstructed image of the skull (see Fig. 1).

Fig. 1. Triangular defect of the hard palate in a 3D reconstructed image of the skull using X-ray computed tomography (arrow).
Рис. 1. Треугольный дефект твердого нёба при 3D-реконструкции черепа на рентгеновской компьютерной томографии (стрелка).

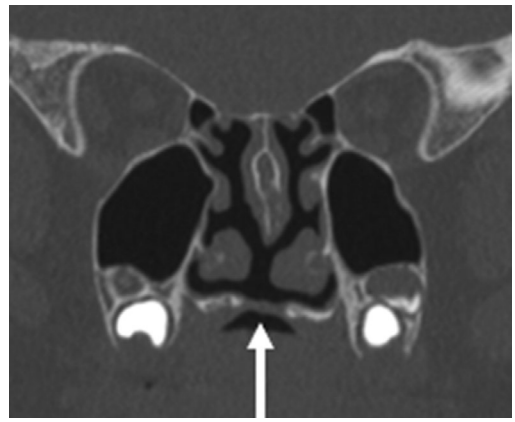

Fig. 2. Defect of the hard palate (arrow) and short vomer in the coronal view in an X-ray computed tomography image.
Рис. 2. Дефект твердого нёба (стрелка) и укорочение сошника в коронарной проекции на рентгеновской компьютерной томографии.

2. In the frontal view, all patients with SMCP involving both the hard and soft palate—and no history of palatoplasty—demonstrated a palatal defect and a shortened vomer that normally combines with the posterior nasal spine and forms choanae (see Fig. 2). However, an isolated vomer shortening on CT may represent its hypoplasia or aplasia and result from surgical intervention by otorhinolaryngologist after endoscopic choanoplasty, transsphenoidal pituitary approaches, and other procedures.
3. In the midsagittal view, anterior displacement of the posterior nasal spine was observed, which subjectively enlarges nasopharynx on CT in the anteroposterior plane. If a hard palate defect is significant, posterior ends of the inferior nasal turbinates protruding into the nasopharynx may be observed (see Fig. 3) [19].

Two patients with characteristic clinical signs of SMCP underwent MRI, where a series of T1- and T2-weighted MR scans revealed a linear hypointense structure along the midline between soft palate muscle fibers, which attests to intermittent levator muscles of the soft palate (see Fig. 4).

Following velopharyngoplasty or palatoplasty, the hypointense structure is no longer observed, as surgical reconstruction of the palatal aponeurosis is performed. Therefore, the combination of hypointense linear structure and intermittent levator muscle contour on MRI may serve as an extra diagnostic criterion for SMCP, in addition to patient history and physical examination findings.

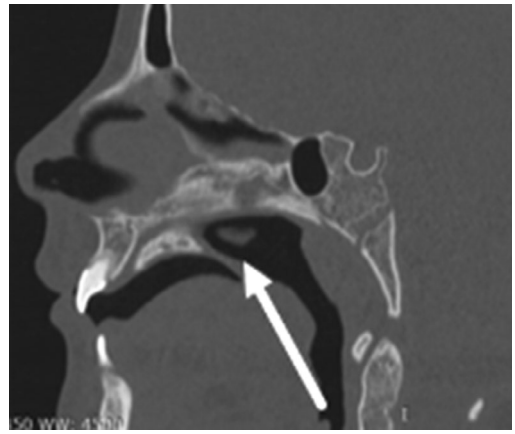

Fig. 3. Anterior displacement of the posterior nasal spine, short vomer and posterior ends of the inferior nasal turbinates protruding into the lumen of the nasopharynx (arrow) in the sagittal view in an X-ray computed tomography image.
Рис. 3. Смещение задней носовой ости кпереди, укороченный сошник и выступающие в просвет носоглотки задние концы нижних носовых раковин (стрелка) в сагиттальной проекции на рентгеновской компьютерной томографии.

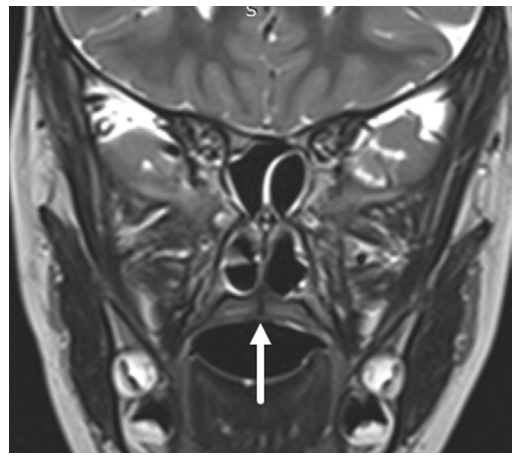

Fig. 4. Hypointense linear structure along the midline of the palate (arrow) in T2 view in a magnetic resonance image.
Рис. 4. Гипоинтенсивная линейная структура по средней линии нёба (стрелка) в Т2-режиме на магнитно-резонансной томографии.

CONCLUSION

Patients seek medical help for upper airways infections from an otorhinolaryngologist much earlier than from a maxillofacial surgeon. Accordingly, an otorhinolaryngologist can detect SMCP manifestations and effects and suspect the defect much earlier than other medical professionals. In case of the compensated SMCP and poor clinical symptoms,

diagnosis without fiberoptic rhinopharyngoscopy, CT, or MRI is challenging. A promising path in SMCP identification is to use radiologic imaging methods. Employing clear diagnostic CT markers and MRI signs in routine practice enables all medical professionals to avoid diagnostic errors. Timely SMCP detection will allow earlier rehabilitation to improve the quality of life and speech.

ADDITIONAL INFORMATION

Author contributions: I.G. Andreeva: conceptualization, methodology, investigation, formal analysis, writing—original draft, literature review; D.I. Marapov: formal analysis; P.V. Tokarev, A.N. Rudyk, E.V. Urakova, R.Yu. Ilyina: writing—review & editing. All authors approved the version of the manuscript to be published, and agreed to be accountable for all aspects of the work, ensuring that questions related to the accuracy or integrity of any part of it are appropriately reviewed and resolved.
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Источники финансирования. Авторы заявляют об отсутствии внешнего финансирования при проведении исследования.
Раскрытие интересов. Авторы заявляют об отсутствии отношений, деятельности и интересов за последние три года, связанных с третьими лицами (коммерческими и некоммерческими), интересы которых могут быть затронуты содержанием статьи.
Оригинальность. При создании настоящей работы авторы не использовали ранее опубликованные сведения (текст, иллюстрации, данные).
Доступ к данным. Все данные, полученные в настоящем исследовании, доступны в статье.
Генеративный искусственный интеллект. При создании настоящей статьи технологии генеративного искусственного интеллекта не использовали.
Рассмотрение и рецензирование. Настоящая работа подана в журнал в инициативном порядке и рассмотрена по обычной процедуре. В рецензировании участвовали два внутренних рецензента.

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