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Contents

Thrice Monthly Volume 11 Number 26 September 16, 2023

MINIREVIEWS

- 6031 Diabetes among Muslims during Ramadan: A narrative review

Ochani RK, Shaikh A, Batra S, Pikale G, Surani S

ORIGINAL ARTICLE

Retrospective Cohort Study

- 6040 Clinical evaluation of ventilation mode on acute exacerbation of chronic obstructive pulmonary disease with respiratory failure

Wang JJ, Zhou Z, Zhang LY

Retrospective Study

- 6051 Predictive value of preoperative albumin-bilirubin score and other risk factors for short-term outcomes after open pancreatoduodenectomy

Zavrtanik H, Cosola D, Badovinac D, Hadžialjević B, Horvat G, Plevel D, Bogoni S, Tarchi P, de Manzini N, Tomažič A

- 6066 Lyophilized recombinant human brain natriuretic peptide for chronic heart failure: Effects on cardiac function and inflammation

Li F, Li H, Luo R, Pei JB, Yu XY

- 6073 Continuous renal replacement therapy with oXiris® in patients with hematologically malignant septic shock: A retrospective study

Wang J, Wei SR, Ding T, Zhang LP, Weng ZH, Cheng M, Zhou Y, Zhang M, Liu FJ, Yan BB, Wang DF, Sun MW, Cheng WX

- 6083 Serum basic fibroblast growth factor and interleukin-1 β predict the effect of first-line chemotherapy in patients with advanced gastric cancer

Zheng L, Gan LH, Yao L, Li B, Huang YQ, Zhang FB, Kuang MQ, Fang N

- 6091 Multinucleated giant cells of bladder mucosa are modified telocytes: Diagnostic and immunohistochemistry algorithm and relation to PD-L1 expression score

Gulinac M, Velikova T, Dikov D

Clinical Trials Study

- 6105 Comparing the efficacy of regen-cov, remdesivir, and favipiravir in reducing invasive mechanical ventilation need in hospitalized COVID-19 patients

Hegazy SK, Tharwat S, Hassan AH

META-ANALYSIS

- 6122 Risk factors for stroke recurrence in young patients with first-ever ischemic stroke: A meta-analysis

Xia Y, Liu H, Zhu R

SCIENTOMETRICS

- 6132** Unveiling the hidden world of gut health: Exploring cutting-edge research through visualizing randomized controlled trials on the gut microbiota

Zyoud SH, Shakhshir M, Abushanab AS, Koni A, Shahwan M, Jairoun AA, Abu Taha A, Al-Jabi SW

CASE REPORT

- 6147** Rivaroxaban for the treatment of heparin-induced thrombocytopenia with thrombosis in a patient undergoing artificial hip arthroplasty: A case report

Ly FF, Li MY, Qu W, Jiang ZS

- 6154** Mepolizumab induced palmoplantar psoriasis: A case report

Artosi F, Diluvio L, Vultaggio M, Campione E, Bianchi L

- 6159** Early diagnosis of renal pelvis villous adenoma: A case report

Li LL, Song PX, Xing DF, Liu K

- 6165** Identification of the dominant loop of a dual-loop macro-reentry left atrial flutter without prior intervention using high-density mapping technology: A case report

Yu SD, Chu YP

- 6170** Surgery for fibrous dysplasia associated with aneurysmal-bone-cyst-like changes in right proximal femur: A case report

Xie LL, Yuan X, Zhu HX, Pu D

- 6176** Efficacy of abatacept treatment in a patient with enteropathy carrying a variant of unsignificance in *CTLA4* gene: A case report

Musabak U, Erdoğan T, Ceylaner S, Özbek E, Suna N, Özdemir BH

- 6183** Postpartum hemophagocytic lymphohistiocytosis: A case report

An JH, Ahn JH

- 6189** Non-arteritic anterior ischemic optic neuropathy combined with branch retinal vein obstruction: A case report

Gong HX, Xie SY

- 6194** Large colonic lipoma with a laterally spreading tumor treated by endoscopic submucosal dissection: A case report

Bae JY, Kim HK, Kim YJ, Kim SW, Lee Y, Ryu CB, Lee MS

- 6200** T/myeloid mixed-phenotype acute leukemia treated with venetoclax and decitabine: A case report

Park S, Jeong EJ, Kang JH, Lee GW, Go SI, Lee DH, Koh EH

- 6206** Severe inflammatory disorder in trisomy 8 without myelodysplastic syndrome and response to methylprednisolone: A case report

Pan FY, Fan HZ, Zhuang SH, Pan LF, Ye XH, Tong HJ

- 6213** Aggressive variant prostate cancer: A case report and literature review
Weng XT, Lin WL, Pan QM, Chen TF, Li SY, Gu CM
- 6223** Typical Zollinger-Ellison syndrome-atypical location of gastrinoma and absence of hypergastrinemia: A case report and review of literature
Zhang JM, Zheng CW, Li XW, Fang ZY, Yu MX, Shen HY, Ji X
- 6231** Left epigastric isolated tumor fed by the inferior phrenic artery diagnosed as ectopic hepatocellular carcinoma: A case report
Liu HB, Zhao LH, Zhang YJ, Li ZF, Li L, Huang QP
- 6240** Squamous cell carcinoma associated with endometriosis in the uterus and ovaries: A case report
Cai Z, Yang GL, Li Q, Zeng L, Li LX, Song YP, Liu FR
- 6246** Intestinal obstruction due to giant liver cyst: A case report
Küçük A, Mohamed SS, Abdi AM, Ali AY
- 6252** Difficulties in diagnosing angiomatoid fibrous histiocytoma of the head and neck region: A case report
Michcik A, Bieñ M, Wojciechowska B, Polcyn A, Garbacewicz Ł, Kowalski J, Drogoszewska B
- 6262** Efficacy of tolvaptan in an infant with syndrome of inappropriate antidiuretic hormone secretion associated with holoprosencephaly: A case report
Mori M, Takeshita S, Nakamura N, Mizuno Y, Tomita A, Aoyama M, Kakita H, Yamada Y
- 6268** Recurrent hemoptysis in pediatric bronchial Dieulafoy's disease with inferior phrenic artery supply: A case report
Wang F, Tang J, Peng M, Huang PJ, Zhao LJ, Zhang YY, Wang T
- 6274** Variant of Guillain-Barré syndrome with anti-sulfatide antibody positivity and spinal cord involvement: A case report
Liu H, Lv HG, Zhang R
- 6280** Secondary pulmonary infection by *Fusarium solani* and *Aspergillus niger* during systemic steroid treatment for COVID-19: A case report
Usuda D, Kato M, Sugawara Y, Shimizu R, Inami T, Tsuge S, Sakurai R, Kawai K, Matsubara S, Tanaka R, Suzuki M, Shimozawa S, Hotchi Y, Osugi I, Katou R, Ito S, Mishima K, Kondo A, Mizuno K, Takami H, Komatsu T, Oba J, Nomura T, Sugita M
- 6289** Collision tumor of primary malignant lymphoma and adenocarcinoma in the colon diagnosed by molecular pathology: A case report and literature review
Jiang M, Yuan XP
- 6298** Successful resolution of gastric perforation caused by a severe complication of pancreatic walled-off necrosis: A case report
Noh BG, Yoon M, Park YM, Seo HI, Kim S, Hong SB, Park JK, Lee MW
- 6304** Bilateral dislocation of the long head of biceps tendon with intact rotator cuff tendon: A case report
Sohn HJ, Cho CH, Kim DH

- 6311** Delayed diagnosis of abdominal Henoch-Schonlein purpura in children: A case report

Guo H, Wang ZL, Tao Z

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Difficulties in diagnosing angiomatoid fibrous histiocytoma of the head and neck region: A case report

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Abstract

BACKGROUND

Angiomatoid fibrous histiocytoma (AFH) is a rare, slow-growing soft tissue tumor. It appears mostly on the limbs and trunk in children and young adults. The biology of AFH remains unclear because of the small number of reported cases. Diagnostic testing does not provide definitive results. It has two clinical forms, that differ in terms of gene expression and clinical prognosis. It is important to inform the laboratory which specific gene testing is necessary. Here, we describe a case of rare AFH in the submandibular region using a full genetic panel.

CASE SUMMARY

A 13-year-old boy who had been misdiagnosed in the past 6 mo by his dentist visited our clinic because of a lesion in the submandibular area on the right side. The lesion was homogeneous and painless upon palpation. No skin discoloration was observed. Due to the non-specific radiological picture computed tomography (CT), magnetic resonance imaging (MRI), cone-beam CT (CBCT), and ultrasound-guided biopsy were performed. A venous malformation was suspected on the MRI. None of the tests provided a definitive diagnosis. Owing to the non-specific radiological findings, the patient qualified for surgical treatment. The surgical procedure included an excisional biopsy. The diagnostic testing was extended using gene rearrangements. The most distinctive gene translocation in diagnosing AFH is within the EWS RNA-binding protein 1 (EWSR1)-CREB-binding protein. However, in this case, the diagnosis was confirmed by a rearrangement within the EWSR1 gene testing.

CONCLUSION

AFH in the submandibular location is rare, and surgical treatment with genetic evaluation defines AFH type that affects subsequent procedures.

Key Words: Angiomatoid fibrous histiocytoma; Head and neck; Fine-needle aspiration; CREB-binding protein; Gene expression; Computed tomography; Case report

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Core Tip: Angiomatoid fibrous histiocytoma (AFH) is a rare neoplasm of soft tissues with an atypical location and a large morphological spectrum, and is considered a lesion with diagnostic difficulties. Auxiliary diagnostic tests in the field of molecular genetics are also important. The EWS RNA-binding protein 1-CREB-binding protein 1 t(2;22)(q33;q12) fusion assay is an extremely important diagnostic element that should be routinely performed in patients with AFH. In the case of gene translocations, we are obliged to increase the frequency of follow-up examinations. However, owing to the small number of clinical cases, it is difficult to predict the course of the disease and assess patient prognosis.

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INTRODUCTION

Angiomatoid fibrous histiocytoma (AFH) is a rare, slow-growing, soft tissue tumor with a low risk of malignancy[1]. It accounts for 0.3% of all soft tissue sarcomas. It was first described in 1979 as “angiomatoid malignant fibrous histiocytoma” and it was classified by the World Health Organization under the category of “tumors of uncertain differentiation” in 2020. AFH is a lesion that rarely metastasizes[2-4]. It mostly occurs on the superficial extremities of both children and young adults[3,5-7]. It can be misdiagnosed because its clinical, histological, and radiological findings are similar to those of several tumors[5,8].

Here, we present the case of a young male with a lesion in the submandibular area on the right side. It is a rare location. In the available literature, AFH lesions have been described in the upper and lower limbs but less often in the head and neck[9]. Because of the rarity of AFH, we also performed a genetic test using the fluorescent in-situ hybridization (FISH) method, which detected a rearrangement of the EWS RNA binding protein 1 (*EWSR1*) gene. Diagnostic gene testing was extended to a specific test for the translocation within the *EWSR1*-CREB-binding protein (*EWSR1*-CREB). No rearrangement was observed within these genes in the present study (Table 1).

CASE PRESENTATION

Chief complaints

A 13-year-old boy was admitted to the hospital because of a lesion in the right submandibular area that had persisted for six months.

History of present illness

The patient was referred from a dentist, and was generally healthy and without allergies.

History of past illness

There was no obvious history of illness related to the disease.

Personal and family history

There was no significant personal or family history related to the disease.

Physical examination

Clinically, the lesion was firm, homogeneous, compact, painless to palpation, and 2.5 cm × 2.0 cm in size. No skin discoloration was observed. The boy did not complain of any sensory disorders and denied any growth changes.

Laboratory examinations

Routine blood test (November 2022) results were as follows: Neutrophil cell count, $3.88 \times 10^9/L$ (reference value: $2-7 \times 10^9$

Table 1 Timeline

Time	Name of procedure
November 2022	Laboratory examinations
June 2022	CT Siemens Somatom Definition Flash (Siemens Healthcare, Germany) with standard contrast enhanced protocol
July 2022	First MRI (1.5T MR Magnetom Aera Flash; Siemens Healthcare, Germany) with contrast injection protocol
October 2022	FNA
November 2022	CBCT
November 2022	First surgical procedure
January 2023	Second MRI (1.5T MR Magnetom Aera Flash; Siemens Healthcare, Germany) with contrast injection protocol
January 2023	Second surgical procedure

CBCT: Cone beam computed tomography; FNA: Fine-needle aspiration; MRI: Magnetic resonance imaging.

/L); neutrophil percentage: 58.6% (reference value: 40%-74%).

Imaging examinations

The first computed tomography (CT) (Siemens Somatom Definition Flash; Siemens Healthcare, Germany) was performed on June 23, 2022. It showed a lesion under the mandible on the right side with smooth edges. It was not connected to the sublingual or submandibular glands. Owing to the nonspecific CT image, magnetic resonance imaging (MRI) (1.5T MR Magnetom Aera Flash; Siemens Healthcare, Germany) with contrast (Gadovist, 5 mL) was performed on July 25, 2022. In the submandibular space on the right side, heterogeneous high signal intensity was observed on T2-weighted images, with a hemorrhagic component measuring 18 mm × 25 mm × 13 mm. The lesion adhered to the digastric muscle and the body of the mandible and involved the platysma (Figure 1).

Conclusion: A venous malformation was suspected. The radiologist suggested extending the diagnosis using ultrasonography to plan the treatment. The large interval between diagnostic imaging and testing is due to the patient's lack of cooperation and not reporting for follow-up to the Department of Maxillofacial Surgery.

Fine-needle aspiration (FNA) was performed on October 27, 2022. In the submandibular space on the right side, visible polycyclic, solid, isoechogenic, and hypoechogenic lesions up to 23 mm × 24 mm × 14 mm were observed. The mass was hard with congenital vascularity. It was not equivalent to lymphatic or lymphatic-venous malformations.

Microscopic results of FNA: The cervical smear included sparse, incoherent extravasation of blood and several crushed parts of tissues. Furthermore, a few cells were similar to each other, such as cells with an oval nucleus and no mitosis. FNA does not reveal the characteristics of the lesion. Surgical excision of the entire lesion was necessary (Figure 2).

Single bland cell with oval nucleus, smooth nuclear membranes, and conspicuous nucleolus.

None of these tests provided a diagnosis, and the patient was eligible for surgery under general anesthesia. The surgical procedure included an excisional biopsy. Before that, we extended the diagnostics with cone beam CT (CBCT) on November 10, 2022, which revealed a 3 mm lesion in the cortical layer of the mandible on the right side (Figure 3).

FURTHER DIAGNOSTIC WORK-UP

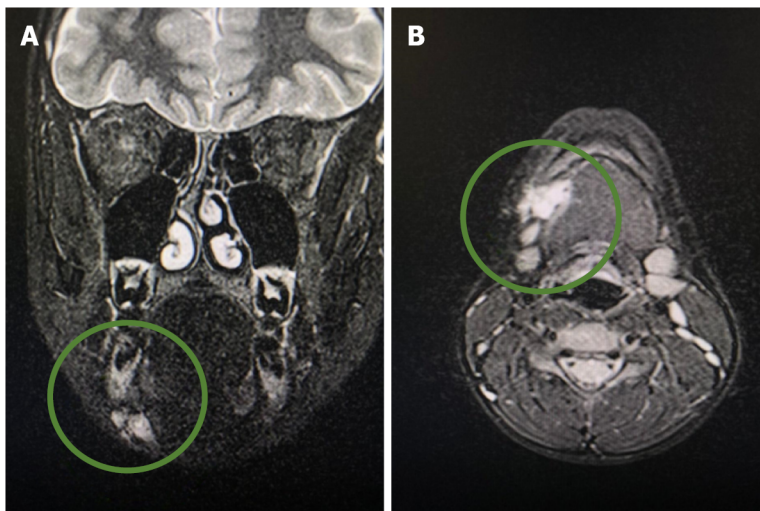
Histopathological diagnosis: Irregular fibrohistiocytic hyperplasia with a variety of cells was observed between fibrous tissue, fatty tissue, and fibrous skeletal muscles. They were surrounded by multiple blood vessels. In addition, minor lymphocytes are rare in this circuit. Hyperplasia is an invasive and expandable condition. The phenotype of hyperplasia is as follows: SMA+, CD68+, CD163+, CD99+, desmine-, beta-catenin-, EMA-, CKAE1/AE3-, S100-, ALK-Bond-, CD34-, ERG-, and indeks Ki-67 IS 6% (Figure 4).

FINAL DIAGNOSIS

The cumulative findings suggested fibrohistiocytic hyperplasia with an intermediate degree of malignancy, AFH.

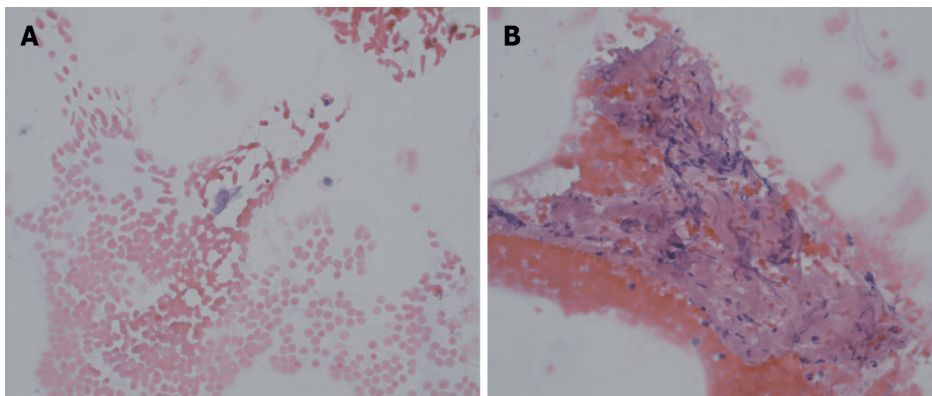
TREATMENT

The surgery was performed on November 29, 2022, under general anesthesia with oral intubation. A skin incision was made on the neck and the mass alone was removed, avoiding macroscopically unchanged tissues, which was considered the best solution. The affected external layer of the bone was exposed during the procedure. Along with the tumor, a bone



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Figure 1 Computed tomography. A: Focal change on the right side of the submandibular space initially diagnosed as a venous malformation; B: Visible irregular outline of a moderately hyperintense area of 18 mm × 25 mm × 13 mm.



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Figure 2 Fine needle aspiration at high magnification (400 ×, hematoxylin and eosin staining). A: Single bland cell with oval nucleus, smooth nuclear membranes, and conspicuous nucleolus; B: Spindle bland cells within an eosinophilic background.

sample was sent for histopathological examination. No complications occurred (Figure 5A and B).

Genetics research

Because of the rarity of AFH, we also performed a genetic test using the FISH method to check for the presence of the genetic translocation, that is, rearrangement gene EWSR1. Rearrangement gene EWSR1 was confirmed. The genetic study was conducted at the Laboratory of the Genetics Clinic of UCK in Gdansk. Pathomorphological and genetic studies validated the diagnosis of AFH.

As we know from previous research, the most aggressive form of AFH is associated with the involvement of both EWSR1 and CREB1 gene rearrangements. Tumors harboring these two genes exhibit recurrence and metastasis. Additional testing was performed to rule out the possibility that the tumor was aggressive.

Results of genetic testing

EDTA- and formalin-fixed, paraffin-embedded samples were submitted for testing. A total of 750 genes were tested, including CREB1, methylthioadenosine phosphorylase gene, tumor protein p53, and MRE11 gene (MRE 11).

The novel allele frequency, which is the frequency at which a mutated allele occurs in the sequencing data, was 100%. The observed frequencies are influenced by the tumor content and copy number alterations and do not directly correlate with the frequency of the variant in the tumor. Somatic alterations were classified according to their functional effect on protein levels into the following categories: inactivating/activating/function altered likely inactivating/activating/function altered unknown and benign.

The detected germline variant c.1867+5G > T; p.,? in gene MRE11 is predicted to cause aberrant splicing and might therefore disrupt protein function. Based on the ACMG criteria, the variant was ranked as a variant of uncertain significance in the germline (Tables 2 and 3).

Table 2 Variant with potential therapeutic relevance of gene *MRE11*

Item	Description
Gene	<i>MRE11</i> (germline)
Functional category	Splice_region
Variant	c.1867+5G > T;P.?
NAF in germline	0.52
Effect on protein function	Probably inactivating
Therapeutic option for discussion in the MTB	PARP inhibitor
Approved by EMA/FDA	EMA & FDA
Approved for current entity	Po

PARP inhibitor: Inhibitor of the enzyme poly ADP ribose polymerase. MTB: Molecular Tumor Boards; EMA: European Medicines Agency; FDA: Food and Drug Administration.

Table 3 Relevant therapeutics for gene *MRE11*

Drug name	Tumor entity	Approval	Approval limited to biomarkers/others	Approval in combination with other drugs
Niraparib, PARP inhibitor	Prostate cancer	EMA	BRCA1, BRCA2	Abiraterone
Olaparib, PARP inhibitor	Fallopian tube carcinoma	EMA	HRD	Bevacizumab
Olaparib, PARP inhibitor	Fallopian tube carcinoma	FDA	HRD	Bevacizumab
Olaparib, PARP inhibitor	Ovarian cancer	EMA	Cancer associated with HRD	Bevacizumab
Olaparib, PARP inhibitor	Ovarian cancer	FDA	HRD	Bevacizumab
Olaparib, PARP inhibitor	Primary peritoneal carcinoma	EMA	Cancer associated with HRD	Bevacizumab
Olaparib, PARP inhibitor	Primary peritoneal carcinoma	FDA	HRD	Bevacizumab
Olaparib, PARP inhibitor	Prostate cancer	EMA	Adult patients, metastatic castration- resistant cancer, chemotherapy not indicated	Abiraterone, prednisone
Olaparib, PARP inhibitor	Prostate cancer	FDA	BRCA1, BRCA2	Abiraterone, prednisone

BRCA1: Breast cancer type 1 susceptibility protein; BRCA2: Breast cancer type 2 susceptibility protein; HRD: Cancer associated with Homologous Recombination Deficiency.

The variant 28/*28 detected in UDP-glucuronosyltransferase family 1 member A1 is a homozygous germline variant. For this genotype [also known as (TA)7/(TA)7rs8175347 or rs3064744], there is a possible increase in toxicity upon treatment with irinotecan-based chemotherapeutics (Table 4).

No other genes or translocations, such as EWSR1-CREB, were found.

Operative technique

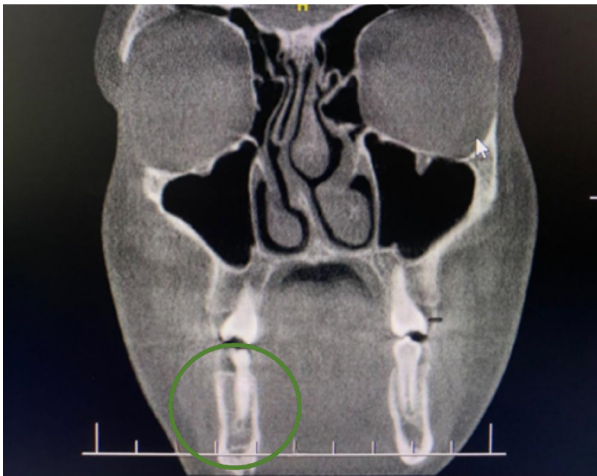
We also followed-up with a new MRI with contrast (Gadovist, 5.5 mL) on January 3, 2023 (Figure 6).

Based on histopathological examination results, a second surgery was conducted to completely remove the lesions with wide margins.

The second surgery was performed on January 24, 2023, under general anesthesia with oral intubation. A skin incision was made within the scar from the initial surgery, and part of the platysma was removed, as well as part of the increased cohesion tissue located under the body of the mandible and surrounding lymph nodes. The specimen was sent for histopathological examination. Intraoperative course was without complications. Intravenous pharmacotherapy administered after the first surgery was repeated (Figure 5C).

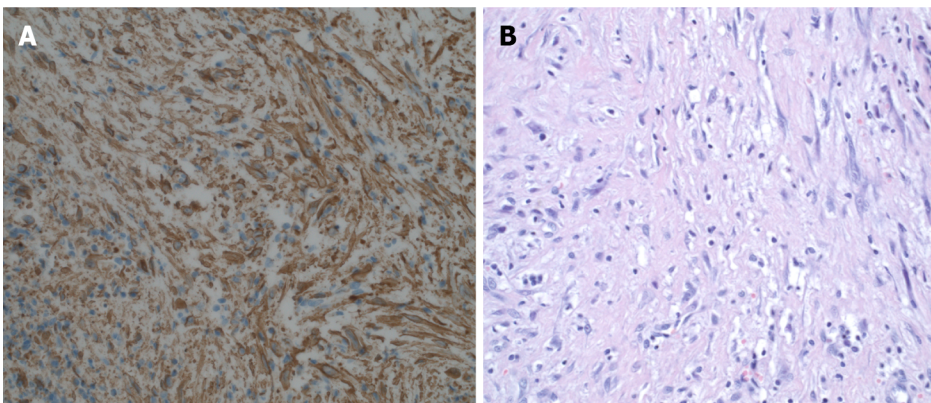
Table 4 Variants with pharmacogenetic relevance gene UDP glucuronosyltransferase family 1 member A1

Item	Description
Gene	UGT1A1
Functional category	Upstream_gene
Variant	c.-41_-40dupATp.?(*28/*28)
Transcript-ID	NM_000463.3
Zygosity in germline	Homozygous
Effect on protein function	Inactivating
Therapeutic option	Topoisomerase inhibitor
Phenotype	Poor metabolizer



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Figure 3 A smooth contoured 3 mm defect in the cortical layer of the inner mandible on the right side near the border of the scan.

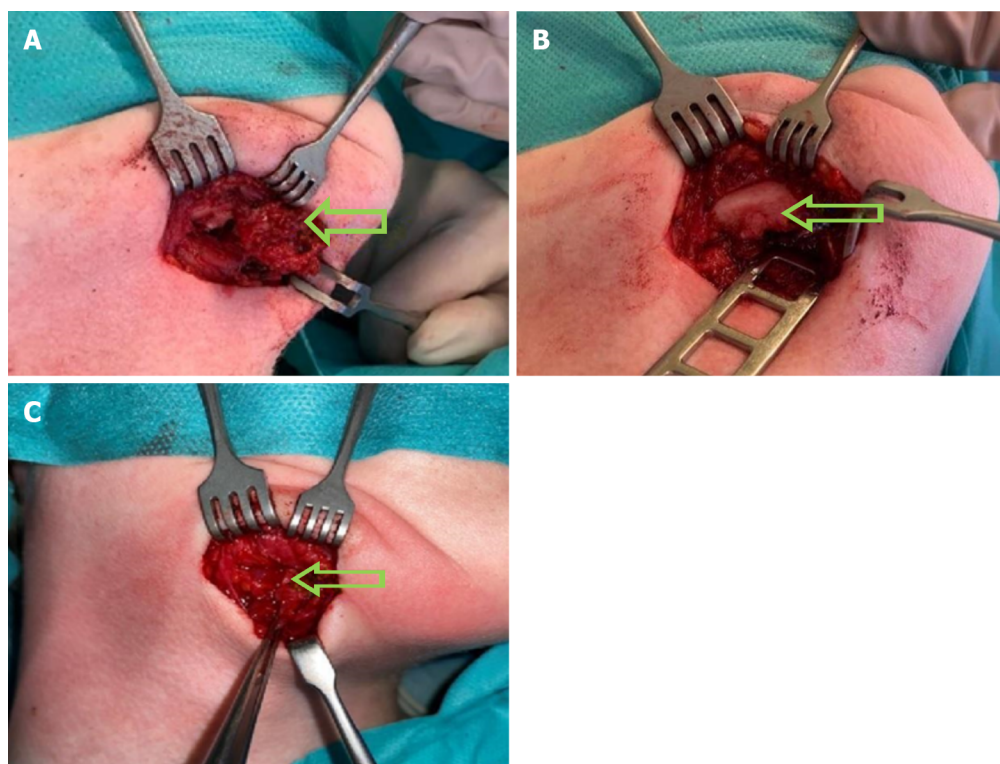


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Figure 4 Histopathological diagnosis. A: Diffuse expression of smooth muscle actin. Excision, magnification 400 ×; B: Excision (400 ×, hematoxylin and eosin staining). Short spindled myoid and ovoid, histiocyte-like cells within a somewhat myxoid background.

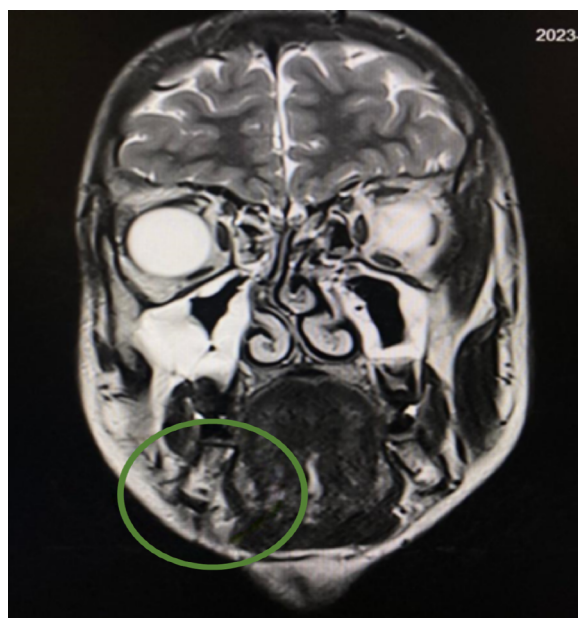
OUTCOME AND FOLLOW-UP

Based on the results of the histopathological examination extended by genetic examination, the presence of an extremely rare soft-tissue sarcoma, AFH, was confirmed. To the best of our knowledge, this is the first reported case involving this location. Since gene translocation has not been demonstrated, particularly in EWSR1-CREB, surgery can be considered the main and final treatment (Figure 7). The patient is currently reporting for routine check-ups at the Oral and Maxillo-facial Surgery Clinic. Locally, there were no signs of recurrence.



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Figure 5 Intraoperative picture. A: Intraoperative picture of submandibular, angiomatoid fibrous histiocytoma, tumor is marked with an arrow; B: Intraoperative picture of a destroyed external layer of the mandible, it is marked with an arrow; C: Intraoperative view of the second procedure, no AFH irregularities were found. Tissue scar are marked with arrows.



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Figure 6 Magnetic resonance imaging before second surgical procedure. Just below the outline of the body of the mandible, a lesion suspected of a residual tumor mass of 4 mm × 8 mm × 4 mm is visible.

DISCUSSION

AFH, first discovered in 1979, is a very rare soft tissue tumor with a relatively good prognosis and medium malignancy potential[10,11]. It accounts for 0.3% of all soft tissue sarcomas. It is considered an intermediate tumor. In the past, it was classified as a malignant variant, but owing to its course and treatment, it was found to have a favorable prognosis[12]. It

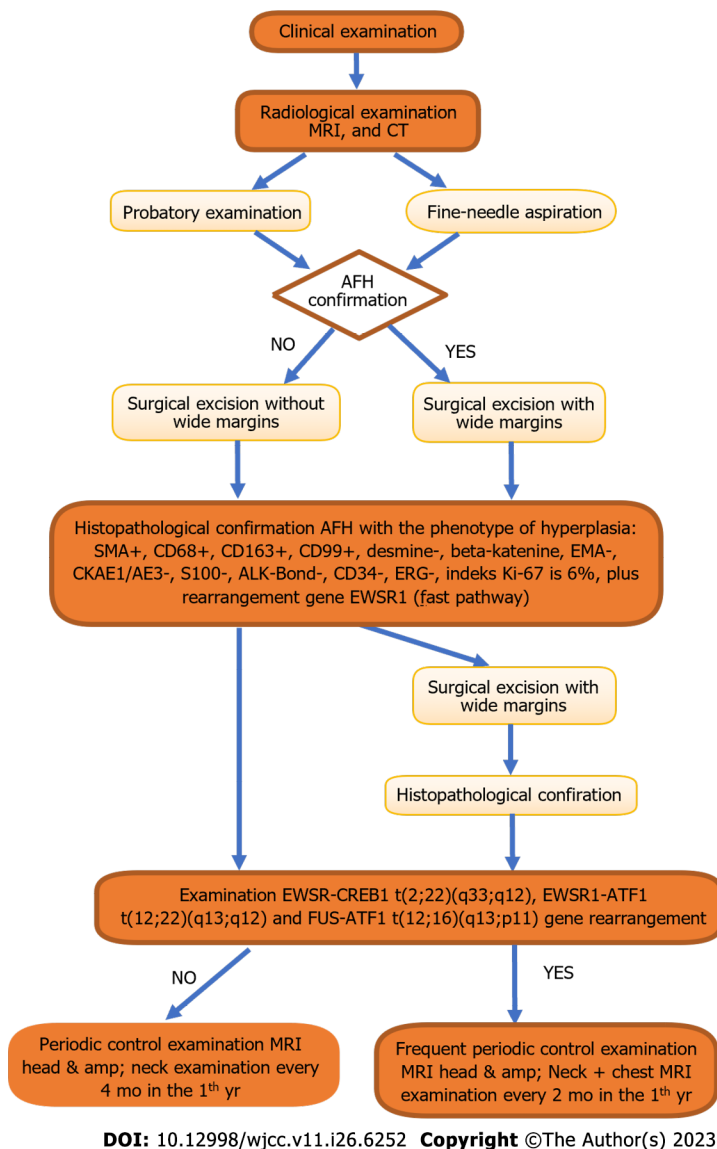


Figure 7 Proposed diagnostic and therapeutic path. AFH: Angiomatoid fibrous histiocytoma; CT: Computed tomography MRI: Magnetic resonance imaging.

occurs most often in the deep dermis and subcutaneous tissues of the upper and lower limbs[6]. It mainly affects children and young adults, and there have been a few reported cases involving older adults[12]. The tumor growth is slow and painless. Patients are often unaware of this disease[2]. This may be accompanied by fever and general weakness. However, this criterion does not define a unit[11].

Based on the case presented, diagnosis is difficult and yields ambiguous results. From the clinical and radiological perspectives, lymphatic malformations, lymphovenous malformations, bone aneurysmal cysts, hematomas, hemangiomas, and lipomas can be suspected[5]. Radiological methods for AFH are not specific, and do not lead to a specific diagnosis.

Surgical treatment alone enabled us to obtain additional information. Based on the available literature, surgery involving the removal of a lesion with a wide margin of macroscopically unchanged tissue is the best solution. In the head and neck area, performing a wide excision is often clinically impossible and is associated with disease recurrence in the future. However, local recurrence of the lesion depends on the size and depth of tissue invasion[11] and occurs in 15% [13] or, according to another source, in 2%-12% of cases[5].

Based on immunohistochemistry results, CD68, CD99, EMA, and wimentyne (EP21) were positive for epithelial membrane antigens, whereas cytokeratin and S100 protein were negative[5,11,12]. However, desmine was positive and negative in two different assays[2,5,12].

Because the diagnosis of AFH can be difficult owing to variable histological features and the lack of highly sensitive immunohistochemical markers, the immunohistochemical markers TLE1 and BCOR have been tested and proven useful. The sensitivity and specificity of TLE1 marker for detecting AFH were 100.0% and 94.4%, respectively. Therefore, TLE1 can be considered as a highly sensitive immunohistochemical marker[14].

In addition, FISH tests can detect gene rearrangements. The three most characteristic AFH translocations are: EWSR-CREB1 t(2;22)(q33;q12), EWSR1-ATF1 t(12;22)(q13;q12), and FUS-ATF1 t(12;16)(q13;p11)[5,15,11,16-18]. The most common translocation is EWSR-CREB1 (90%)[19,20]. This is associated with the risk of distant metastases. Therefore, we decided to

keep the patient under observation and scheduled follow-up visits. Distant metastases occur in 5% of cases and are mainly located in the regional lymph nodes or lungs[13]. The EWSR1-CREB fusion is also found in gastrointestinal clear cell sarcoma and primary myxoid lung sarcoma (PPMS)[19]. CCSGT is a rare cancer that occurs in the walls of the small intestine, mainly in young adults and is similar to AFH. In contrast, PPMS is a rare lung cancer that arises intrabronchial [19].

The histology of AFH consists of a multinodular growth of spindle-shaped cells. These cells have a characteristic syncytial appearance. The pseudovascular spaces are filled with cancer cells and blood. Histologically, a thick, fibrous pseudocapsule with deposited hemosiderin together with lymphoplasmacytic cuffs can also be distinguished[21]. Regarding the morphology, AFH showed a wide spectrum. Multinodular cell growth was consistent. A specific feature of the morphology is the presence of small, clear cells with scanty cytoplasm. This resembles Ewing's Sarcoma[21].

Ultrasonography is the least invasive radiological examination. The image shows a heterogeneous hypoechoic structure that may contain cystic spaces or fluid levels[6,19]. On CT, AFH is described as a heterogeneous soft tissue lesion with hyperdense areas suggestive of hemorrhage and hypodense areas that could indicate necrosis or a cystic/myxoid lesion[6]. MRI may show hypointense pseudocapsules (double-rim sign), cystic changes, peritumoral edema, and fluid-fluid levels. On T1-weighted images, the lesion is most often isointense to the muscles, whereas on T2-weighted images, it is heterogeneously hyperintense[6,19,21,22].

Favorable prognosis has been established for patients with AFH after treatment. As mentioned above, local recurrence occurs in up to 15% of cases, whereas distant metastases occur in < 5% of cases. The presence of gene translocations, especially EWSR1-CREB, is also important because of the increased risk of distant metastases. Patients with distant metastases have been treated with systemic chemotherapy; however, no benefits have been reported. AFH has an unspecified course that is rarely predictable[21].

CONCLUSION

AFH is a rare neoplasm of soft tissues with an atypical location and a large morphological spectrum, and is considered a lesion with diagnostic difficulties. In the case of AFH, diagnostics are crucial because they allow us to plan further diagnostics and extend the treatment to distant tumors, in addition to local treatment (in the case of metastases). Auxiliary diagnostic tests in the field of molecular genetics (reverse transcription polymerase chain reaction or FISH) are also important. The EWSR1-CREB1 t(2;22)(q33;q12) fusion assay is an extremely important diagnostic element that should be routinely performed in patients with AFH. In the case of gene translocation, we are obliged to increase the frequency of follow-up examinations, including chest CT imaging. However, owing to the small number of clinical cases, it is difficult to clearly predict the course of the disease and assess the prognosis of patients. Publishing the treatment results for patients with AFH will deepen our knowledge about this tumor.

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