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Novel and unusual *USP6* fusion partners in aneurysmal bone cyst and their role in pathogenesis and histopathological evaluation of this disease

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ABSTRACT

Aims The purpose of this study is to report novel and unusual *USP6* fusion partners in aneurysmal bone cysts (ABCs). These findings may be useful in routine diagnostics as well as in studying the biology of *USP6*-related disorders.

Methods A cohort of seven patients diagnosed with ABC examined between 2014 and 2023 at Motol University Hospital in Prague was included into this retrospective non-randomised study. All cases were analysed using histopathological evaluation, immunohistochemistry and Anchored multiplex RNA methods. Demographic characteristics and clinical data were also analysed.

Results We identified two novel (*ZFX* and *IP6K2*), three unusual (*MEF2A*, *EIF1* and *COL1A2*) and two common (*CDH11*) fusion partners with *USP6* gene among all seven cases of ABC.

Conclusions Cases in our study were diagnosed as ABCs due to characteristic clinical and morphological presentation. However, not all cases are as self-evident, and molecular testing is necessary. The identification of these gene alterations can be useful in distinction between true ABC and ABC-like changes among many benign and malignant bone tumours.

INTRODUCTION

Aneurysmal bone cyst (ABC) belongs to a heterogeneous group of reactive and neoplastic processes collectively known as giant cell-rich lesions of bone. The biological behaviour of these diseases varies from benign reactive lesions to malignant and aggressive neoplasms and their histopathological diagnosis can often be very challenging. Morphological features alone rarely suffice and clinical data or molecular analysis is required.^{1–3} The use of genetic testing in routine practice is helpful only in select cases to date, with ABC being one of them.

A useful diagnostic clue represents the fact that approximately 70% of ABC harbour rearrangements of *USP6* gene, and therefore represent true neoplasms. Such rearrangements are not found in ABC-like changes seen in other tumours.⁴ Plenty of possible fusion partners have been described^{5–7} and are thought to drive neoplasia via promoter swapping mechanism.⁸ We present seven cases of ABC with confirmed *USP6* rearrangements, which revealed two common, three rare and two novel *USP6* fusion partners.

WHAT IS ALREADY KNOWN ON THIS TOPIC

⇒ *USP6* rearrangements are, among other shared features, characteristic for so called 'transient neoplasms'. A recent analysis of *USP6* fusion partners shows that most of these genes are physiologically expressed in some stage of tissue repair and there seems to be evolving a new concept of tissue repair-associated neoplasms.

WHAT THIS STUDY ADDS

⇒ We present two novel and three unusual fusion partners in aneurysmal bone cysts and discuss their role in the pathogenesis of *USP6*-related neoplasms in light of the recent discoveries.

HOW THIS STUDY MIGHT AFFECT RESEARCH, PRACTICE OR POLICY

⇒ Our findings add certainty into routine histopathological practice and encourage further research of the biology of *USP6*-related neoplasms.

MATERIALS AND METHODS

Study cohort

A cohort of seven patients diagnosed with ABC diagnosed between 2014 and 2023 at Motol University Hospital in Prague, Czech Republic, was included into this retrospective non-randomised study. Next to the standard histopathological evaluation, all cases were analysed immunohistochemically and using Anchored multiplex RNA methods. Furthermore, the patient's main demographic characteristics and clinical data were collected and analysed.

Histopathological examination

Biopsy samples were fixed in neutral buffered 4% formaldehyde, transported to the histopathological laboratory, postfixed and embedded in paraffin. If needed, decalcification was performed using chelating agent. Subsequently, the paraffin blocks were sectioned into 4 µm thick histological sections and stained with H&E.

Immunohistochemistry

All biopsies were immunostained for Ki-67 to evaluate proliferation of the neoplastic spindle cells. Thin histological sections (3 µm thick) were used, and each sample was stained using the following



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Table 1 Patient characteristics and clinical findings

Number	Sex	Age (years)	Site	Diagnosis	Microscopy	USP6 fusion partner	Ki-67
1	F	10–15	Clavicle	ABC	Fibrous tissue with foci of calcified osteoid. Pseudocystic spaces without inner lining. Areas of hypercellular tissue with giant multinucleated cells and iron pigment.	CDH11::USP6	2%
2	M	5–10	Mandible	ABC	Short fascicles of elongated cells with oval nuclei (no atypia). Varying number of giant multinucleated cells. Local extravasation of erythrocytes. Bone trabeculae predominantly around margins. Area of adjacent skeletal muscle infiltration.	CDH11::USP6	5%
3	F	40–45	Distal phalanx of right-hand finger	ABC - solid variant	Proliferation of short spindle cells without atypia. Plentiful admixture of giant multinucleated osteoclast-like cells. Abundant vascularisation present. No distinct pseudocystic spaces nor haemosiderin deposits.	EIF1::USP6	15%
4	M	50–55	Distal phalanx of left-hand finger	ABC	Subungual mass of polypoid tumour with pseudocystic changes. Mononuclear spindle cells with bland nuclei and plenty of multinucleated giant osteoclast-like cells. Areas of newly formed woven bone with small deposits of haemosiderin. Pseudocystic spaces are focally filled with blood. Exulceration of polypoid convexity with fibrin and polymorphonuclear cells at base with granular tissue formation. No nuclear atypia.	ZFX::USP6	10%
5	F	25–30	Rib	ABC	Lamellar bone tissue and fragments of fibroadipose tissue with areas of spindle cells without atypia. Giant multinucleated cells are also present.	COL1A2::USP6	5%
6	M	15–20	C7 (posterior elements and body)	ABC	Uniform population of oval mononuclear cells and giant osteoclast-like elements with excess nuclei. Mitotic activity visible, but no atypical mitoses present. Areas of collagen-rich stroma. Short fascicles of spindle cells. Pseudocystic spaces engorged with blood in the centre of the lesion. Septa contain immature osteoid. Periphery of lesion with regressive changes of bone and cartilaginous tissue.	MEF2A::USP6	10%
7	F	10–15	C4 (dorsal part)	ABC	Solid and partially cystic lesion consisting of uniform population of oval mononuclear cells without atypia and disperse giant osteoclast-like elements. Pseudocystic spaces are filled with blood and formation of new bone detected at the periphery of the lesion.	IP6K2::USP6	2%

ABC, aneurysmal bone cyst; F, female; M, male.

antibody and protocol: anti-Ki-67 mouse monoclonal antibodies (clone MIB-1, BioSB, dilution 1:150; pre treatment: heating up to 99°C in a pH 6 buffer in a water bath). The detection was performed using a one-step micropolymeric non-biotin system (BioSB, Santa Barbara, California, USA) with a peroxidase complex and 3,3'-diaminobenzidine tetrahydrochloride. The nuclei were counterstained with haematoxylin. To objectively

characterise tumour growth, we counted general proliferation of the neoplastic cells within the whole sample using percentages.

Anchored multiplex RNA

To identify molecular alteration of *USP6* gene and finding its fusion partners, next-generation sequencing was performed.

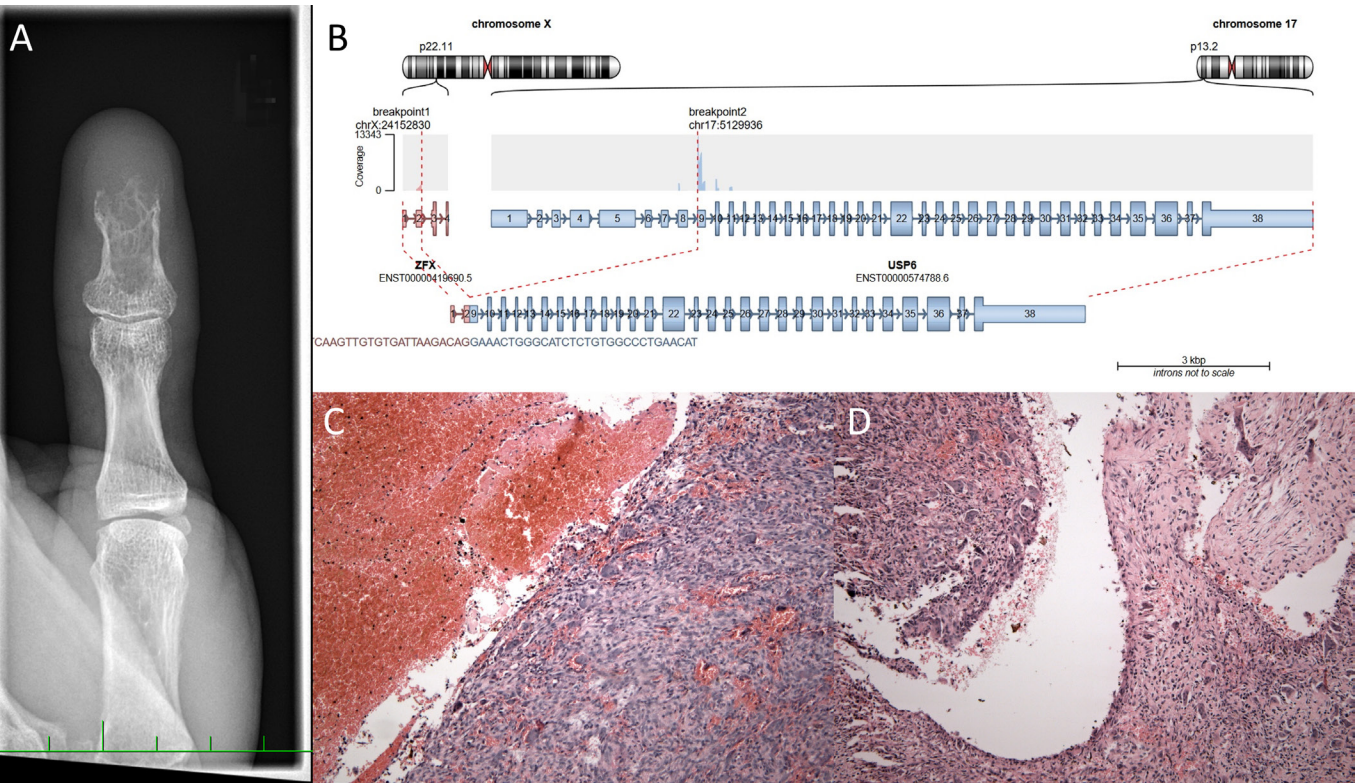


Figure 1 X-ray, microscopy and molecular finding of the case with *ZFX::USP6* gene fusion. (A) X-ray of distal phalanx of the left-hand finger with an osteolytic lesion. (B) Schematic visualisation of a novel detected fusion transcript *ZFX::USP6* using Arriba software (<https://github.com/suhrig/arriba/>). (C,D) Subungual mass of polypoid tumour with pseudocystic changes. The lesion consists of mononuclear spindle cells with bland nuclei and plenty of multinucleated giant osteoclast-like cells. Pseudocystic spaces are focally filled with blood. There is no nuclear atypia detected.

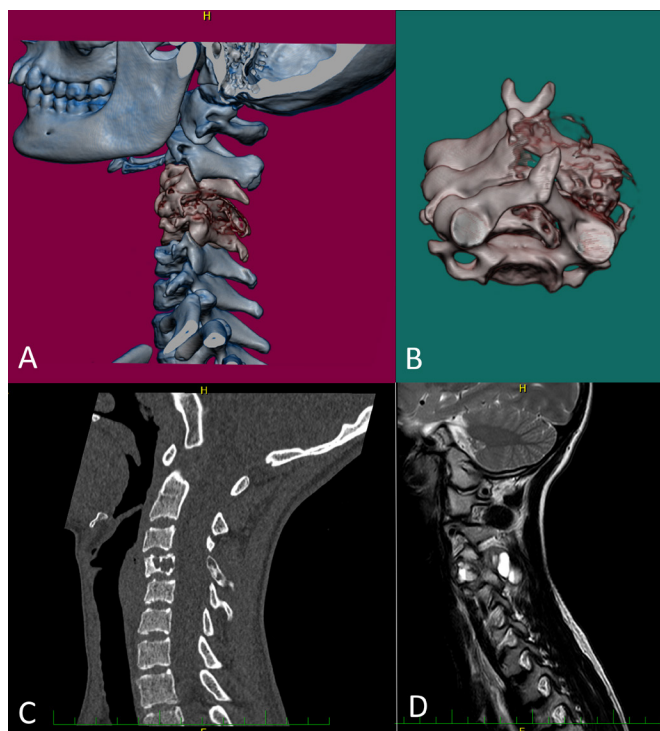


Figure 2 Imaging methods showing aneurysmal bone cyst (ABC) case with *IP6K2::USP6* gene fusion. (A) Three-dimensional (3D) reconstruction of CT scan showing the vertebra C4 involvement, side view. (B) 3D reconstruction of CT scan showing the C4 vertebra involvement, top view. (C) CT showing osteolytic lesion of the vertebra C4. (D) MRI showing a heterogenic mass in the vertebra C4.

The Sarcoma FusionPlex panel (Archer) was used according to the manufacturer's instruction. In brief, RNA was extracted from FFPE (formalin-fixed, paraffin-embedded) sections followed by cDNA synthesis and library preparation. Anchored Multiplex PCR amplicons were sequenced on Illumina MiSeq, and the data were analysed using the Archer and Arriba software.

RESULTS

Clinical features

Seven cases of ABC with confirmed *USP6* rearrangements were analysed in total, four of which were female (57%) and three were male (43%). The mean age was 24.6 years. The lesions were localised within the clavicle, mandible, distal phalanx of the right-hand digit (solid variant of ABC), distal phalanx of the left-hand digit, rib, posterior elements and body of vertebra C7 and dorsal portion of vertebra C4. For more details on patient characteristics, see [table 1](#).

Histological findings and immunohistochemistry

All seven cases showed characteristic histopathological signs of ABC. One case was further subclassified as a solid variant of ABC. All samples consisted of fascicular proliferation of mononuclear bland spindle cells with an admixture of giant multinucleated osteoclast-like cells. Except from the solid variant, there were pseudocystic spaces filled with blood. Proliferation index (Ki-67 positivity) fluctuated around 2%–15% in general with sparse mitotic activity in some cases. However, no atypical mitoses were found.

The histopathological appearance of cases with novel fusions did not stand out among other cases.

ZFX was identified in the case of distal phalanx of the left-hand involvement in man in early 50s. The lesion presented as a subungual mass of polypoid tumour with pseudocystic changes, which consisted of mononuclear spindle cells with bland nuclei and plenty of multinucleated giant osteoclast-like cells. Areas of newly formed woven bone with small deposits of haemosiderin were also detected. Pseudocystic spaces were focally filled with blood. The surface of the lesion was exulcerated and covered with fibrin and neutrophils. At the base of the ulcer, there was granular tissue formation. No marked nuclear atypia was present ([figure 1](#)).

Inositol hexakisphosphate kinases (*IP6K2*) was identified in case of dorsal portion of the vertebra C4 involvement in early adolescent patient ([figure 2](#)). The lesion represented a solid and partially cystic mass consisting of uniform population of oval mononuclear cells without atypia and sparse giant osteoclast-like elements. Pseudocystic spaces were filled with blood, and there was a formation of new bone detected at the periphery of the lesion ([figure 3](#)).

Molecular findings

USP6 rearrangements were found in all seven studied cases. Two showed typical *CDH11::USP6* fusion. Another less common *EIF1::USP6* fusion was present in the case of solid variant of ABC within distal phalanx of the right-hand finger. A rare *MEF2A::USP6* fusion was found in the case of C7 vertebral lesion. One unusual *COL1A2::USP6* fusion was identified, although a similar fusion partner *COL1A1* is common. Finally, a novel *ZFX::USP6* fusion was revealed in the case of distal phalanx of the left-hand thumb involvement in a man in the early 50s ([figure 1](#)). Another novel *IP6K2::USP6* fusion was identified in the case of dorsal part of the vertebra C4 involvement in an early adolescent girl ([figure 3](#)).

DISCUSSION

It should be noted that *USP6* gene rearrangements are characteristically found in other benign mesenchymal neoplasms besides ABC, namely in nodular fasciitis (NF), myositis ossificans and fibro-osseous tumour of digits.^{9 10} All these neoplasms share some morphological and clinical features, including a possibility of spontaneous regression and traumatic involvement.¹¹ These similarities are especially apparent in ABC and NF and some authors suggest that these entities represent clonal neoplastic disorders that may belong to the same biologic spectrum.^{9 12} It was also proposed to group these entities together as *USP6* Associated Neoplasms (UANs) to highlight the shared features.⁹ The label 'transient neoplasms' represents another umbrella term which was proposed, as spontaneous regression was reported in some cases of UANs.^{13–16}

Ever since *USP6* rearrangement was discovered in ABC and, soon after, in the remaining above-mentioned entities, more than 50 different *USP6* fusion partners have been reported. Some of them are common to all these neoplasms, some have been associated only with one entity. The list expands as new gene rearrangements are being discovered.¹⁰ A recent study brought valuable insights into the biology of these lesions by examining the role of the *USP6*-related fusion partners in physiological processes. In-depth analysis shows that many of these fusion partners are genes expressed in different phases of tissue repair.¹¹ This suggests that all the above-mentioned neoplasms are, in fact, related and there seems to be emerging concept of 'tissue repair-associated neoplasms'. We hypothesise that the promoter swapping mechanism creates a dormant, susceptible cell which, when introduced into a tissue repair-like microenvironment where the expression of its respective fusion partner gene would physiologically occur, starts the proliferative neoplastic process.⁸

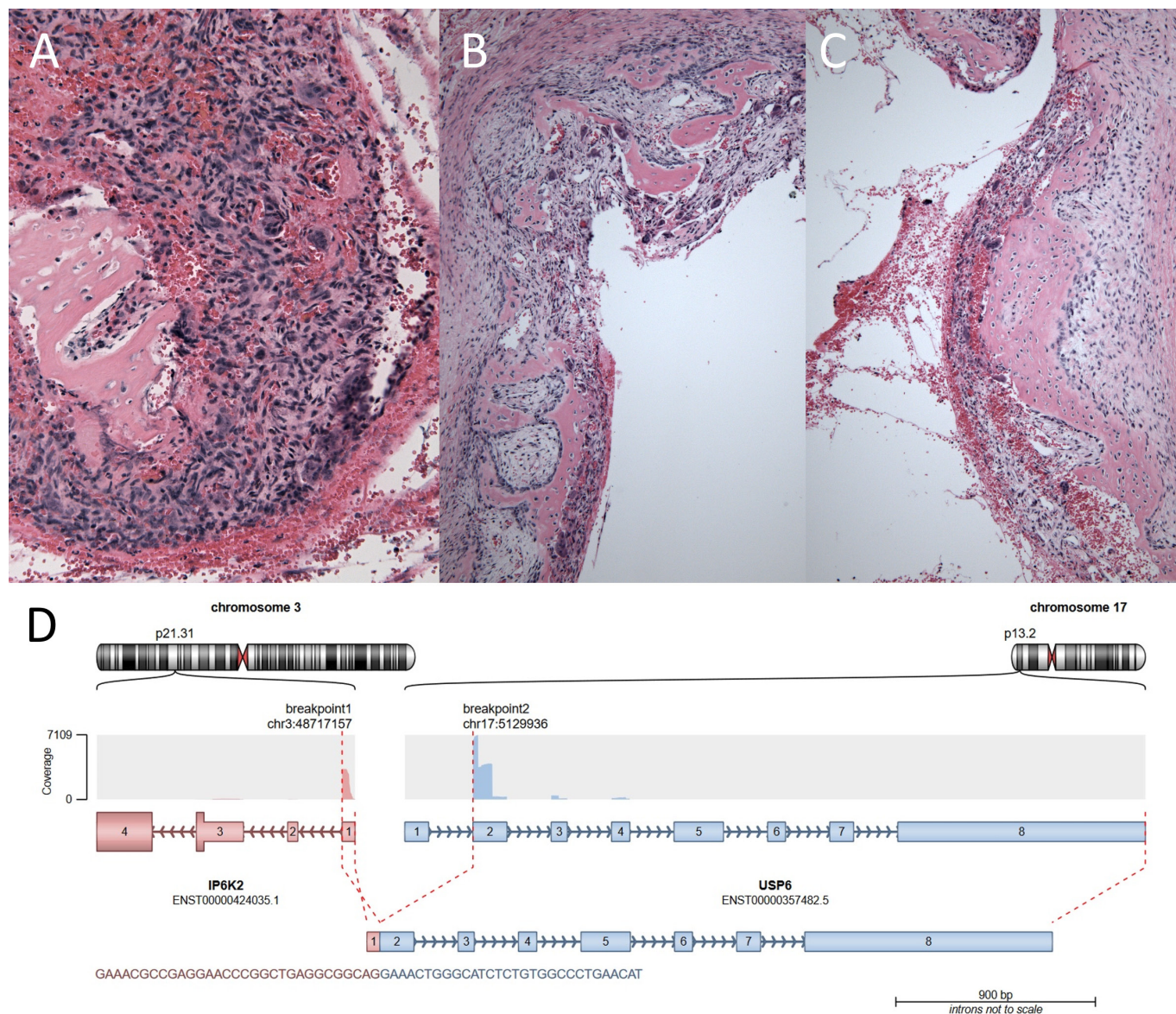


Figure 3 Histopathological and molecular findings of the case with *IP6K2::USP6* gene fusion. (A–C) Solid and partially cystic lesion consisting of uniform population of oval mononuclear cells without atypia and disperse giant osteoclast-like elements. Pseudocystic spaces are filled with blood and formation of new bone can be detected at the periphery of the lesion. (D) Schematic visualisation of a novel detected fusion transcript *IP6K2::USP6* using Arriba software (<https://github.com/suhrig/arriba/>).

Herein, we discuss the role of genes identified in our study as well as review of the literature regarding their involvement in reparation of tissues.

In our study, the most common *CDH11::USP6* fusion in ABC was detected in two of seven (29%) cases; the remaining specimens harboured rearrangements with less common fusion partners^{5–7} and two novel fusion partners. *CDH11* promotes cell proliferation by increasing the sensitivity of cells to platelet-derived growth factor beta and functions as a strong promoter of tissue regeneration.^{17 18} A recent evidence suggests that *CDH11* is also a central mediator of tissue fibrosis which shares similar cellular and molecular pathways with wound healing and tissue repair.¹⁹ *ZFX* gene works as a zinc finger transcription factor physiologically required for self-renewal of haematopoietic stem cells, embryonic stem cells²⁰ and survival of B lymphocytes.²¹ Due to series of ex vivo and in vivo experiments, it has been shown that *ZFX* also plays an important role in skin wound healing by regulating the proliferation and migration

of keratinocytes.²² *IP6K2* gene was identified in the case of dorsal part of the vertebra C4 involvement in early adolescent girl. *IP6K2* belongs to a family of *IP6Ks*—enzymes responsible for creation of high-energy metabolites used in many key biological processes, from regulating telomere length and controlling vesicular trafficking to DNA repair.^{23 24} *MEF2A* gene is highly expressed in many tissues and plays an important role in several biological processes, such as growth, differentiation, survival and neuronal development.²⁵ *MEF2A* involvement in fibroblast activation and hypertrophic scar formation was also described.²⁶ Hypertrophic scars and keloids are the result of an abnormal wound-healing process impaired by inflammation or tension in the lesion.²⁷ *EIF1* gene encodes a protein belonging to a broad range of eukaryotic translation initiation factors. The rate-limiting step of translation is initiation, where *EIF1* plays a crucial role in start codon selection.²⁸ *EIF1* gene is one of the known *USP6*-fusion partners involved in the pathogenesis of ABC.^{29 30} Human type I collagen, which is a major component

of extracellular matrix proteins, is composed of two $\alpha 1$ chains and one $\alpha 2$ chain, encoded by *COL1A1* and *COL1A2*, respectively.³¹ *COL1A2* as a fusion partner can be found in some neoplasms such as in a lipoblastoma, where *COL1A2::PLAG1* rearrangement is characteristic.^{32,33} *COL1A2* fusion with *USP6* was also reported, although a similar fusion partner, *COL1A1*, is much more common in this instance.^{10,34} Moreover, bone marrow-derived fibroblasts make a substantial contribution to the formation of new connective tissue, including type I collagen, during wound repair.³⁵

The majority of fusion partners reported in our study have indeed, according to literature, association with tissue repair, and we suggest that further exploration of this hypothesis may be desirable. Cases in our study were diagnosed as ABCs prior to discovering *USP6* rearrangements due to characteristic clinical and morphological presentation. However, not all cases of ABC are as self-evident and molecular testing is necessary. A considerable number of other bone tumours can morphologically mimic ABC, especially in cases with ABC-like changes, which can sometimes be of severe extent and even obscure the true nature of the original disease, especially in limited samples. The contribution of newly discovered *USP6* fusion partners to routine diagnostics is limited, nevertheless relevant and interesting for understanding the biology of such lesions.

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Competing interests None declared.

Patient consent for publication Not applicable.

Ethics approval This study involves human participants. The present study was approved by the ethics committee of the University Hospital Motol (reference no. EK—771/23) and adhered to the tenets of the Declaration of Helsinki. This is a retrospective study based on tissue material with untraceable patients, therefore the study was properly anonymised.

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