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Unusual fusion gene rearrangements in patients with nodular fasciitis: a study of rare and novel USP6 fusion partners with a review of the literature

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ABSTRACT

Aims This retrospective non-randomised study aims to identify new and rare fusion partners with *USP6* in the setting of nodular fasciitis. It has been proven, that nodular fasciitis can harbour different variants of *USP6* fusions, which can be used in routine diagnostics and even determine the biological behaviour of the process.

Methods A total of 19 cases of nodular fasciitis examined between 2011 and 2022 at Motol University Hospital in Prague were included into this study. Next to the histopathological evaluation, all cases were assessed using immunohistochemistry, RT-PCR and Anchored multiplex RNA methods. Patient's main demographic characteristics and corresponding clinical data were also analysed.

Results This study presents one novel (*KIF1A*) and five rare examples (*TMP4, SPARC, EIF5A, MIR22HG, COL1A2*) of fusion partners with *USP6* among 19 cases of nodular fasciitis.

Conclusion Identification of *USP6* fusion partners in nodular fasciitis helps to understand the biology of such lesions. Moreover, it can be useful in routine histopathological practice of soft-tissues diagnostics, especially in preventing possible misdiagnosis of malignancy.

WHAT IS ALREADY KNOWN ON THIS TOPIC

⇒ In recent years, the detection of various fusion partners with *USP6* has been very popular. However, the literature on majority of them is scarce and limited to isolated case reports.

WHAT THIS STUDY ADDS

⇒ This study presents one novel (*KIF1A*) and five rare examples (*TMP4, SPARC, EIF5A, MIR22, COL1A2*) of fusion partners with *USP6* among 19 cases of nodular fasciitis.

HOW THIS STUDY MIGHT AFFECT RESEARCH, PRACTICE OR POLICY

⇒ Identification of *USP6* fusion partners in nodular fasciitis helps to understand the biology of such lesions. Moreover, it can be useful in routine histopathological practice of soft-tissues diagnostics, especially in preventing possible misdiagnosis of malignancy.

INTRODUCTION

Nodular fasciitis is widely considered to be a benign myofibroblastic neoplasm arising among all age groups but mostly between 20 and 40 years of life.[1–3] The typical localisation tends to be a surface of the fascia of the upper extremities and the head and trunk. However, previous studies indicate that basically any anatomical locality can be involved, including deeper sites.[4–7] Previously disputed neoplastic origin of nodular fasciitis has been confirmed by findings of *USP6* gene rearrangements in the majority of cases.[8–11] Furthermore, *MYH9* gene was identified as the most common *USP6* fusion partner.[6–8] The *USP6* gene rearrangement detection has also been proven as a useful diagnostic tool in cases with morphological uncertainty.[3 7 11]

In recent years, the detection of other fusion partners with *USP6* and their clinical correlations in nodular fasciitis has been very popular and *USP6* fusions with *TPM4*, *EIF5A*, *PPP6R3*, *CTNNB*, *SPARC*, *THBS2*, *COL6A2*, *TNC*, *SEC31A*, *COL1A1*, *COL1A2*, *COL3A1*, *CALU*, *NACA*, *SLFN11*, *LDHA*, *SERPINH1*, *PDLIM7*, *MYL12A*, *PAFAH1B1* and *MIR22HG* have been described in previous studies.[9 12–25] However, the literature on majority of them is scarce and limited to isolated case reports.[9 12–15 18 20–22 25] Furthermore, several publications from recent years have documented peculiar and even malignant behaviour of nodular fasciitis associated with certain fusion partners of *USP6*, which has been recognised by WHO.[15 19] Such findings highlight the importance of recognition and description of new *USP6* rearrangements. Moreover, even cases of nodular fasciitis with typical *MYH9::USP6* fusion can morphologically mimic sarcomatous growth and be easily misdiagnosed as malignancy. Therefore, it is often necessary to assess *USP6* rearrangement to confirm the diagnosis of nodular fasciitis.

This study presents 19 cases of nodular fasciitis examined using histopathology, immunohistochemistry, RT-PCR and Anchored multiplex RNA methods, which revealed one novel and five rare examples of fusion partners with *USP6*.

MATERIALS AND METHODS

A total of 19 cases of nodular fasciitis examined between 2011 and 2022 at Motol University Hospital in Prague were included in this retrospective non-randomised study. Next to the standard histopathological evaluation, all cases were examined using immunohistochemistry, RT-PCR and Anchored multiplex RNA methods. Furthermore,

the patient's main demographic characteristics and corresponding 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. Subsequently, the paraffin blocks were sectioned into 4 µm thick histological sections and stained with H&E.

Immunohistochemistry

All biopsies were immunostained for smooth muscle actin, H-caldesmon, desmin, S100-β and proliferation marker Ki-67, particularly to characterise neoplastic spindle cells. Thin histological sections (3 µm thick) were used, and each sample was stained using the following antibodies and protocols: anti-smooth muscle actin (SMA) mouse monoclonal antibodies (clone 1A4, BioSB— Bioscience for the World, dilution 1:75; pretreatment: heating up to 99°C in a pH 6 buffer in a water bath); anti-H-caldesmon mouse monoclonal antibodies (clone BSB-19|, BioSB, dilution 1:100; pretreatment: heating up to 99°C in a pH 9 buffer in a water bath); anti-desmin mouse monoclonal antibodies (clone D33, BioSB, dilution 1:100; pretreatment: heating up to 99°C in a pH 9 buffer in a water bath); anti-S100-β rabbit monoclonal antibodies (clone EP32, BioSB, dilution 1:300; pre-treatment: heating up to 99 ◦C in a pH 9 buffer in a water bath); anti-Ki-67 mouse monoclonal antibodies (clone MIB-1, BioSB, dilution 1:150; pretreatment: 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.

RT-PCR (detection of MYH9::USP6)

The complementary DNA (cDNA) was synthesised using MMLV Reverse Transcriptase (Invitrogen) from 10 µL total mRNA in a volume of 20 µL. RT-PCR was performed using ×2 PCRBIO HS Taq Mix Red (PCR Biosystems, London, UK) with primers reported previously.[8]

Amplification of a 208 bp amplicon of an abl housekeeping gene was used to confirm the presence of intact and amplifiable cDNA. Direct Sanger sequencing was performed using Big Dye Terminator V.3.1 chemistry (Life Technologies) on positive cases.

Anchored multiplex RNA

Due to the negativity of the *MYH9::USP6* fusion, next-generation sequencin was performed to identify a molecular alteration of *USP6* gene. The Sarcoma FusionPlex panel (Archer) was used according to manufacturer's instruction. In brief, RNA was extracted from tissue cryosections or FFPE 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

Nineteen cases were analysed in total, 11 of which (58%) were female and 8 were male (42%). The median age was 30 years, (range 6 months to 64 years). Sixteen cases (84%) were

Table 1 Patient characteristics and clinical findings in *USP6::MYH9* fusion cases

No.	Sex	Age (years)	Site	Diagnosis	Microscopy	IHC positivity	*USP6* fusion partner
1	F	30–35	Superficial (nuchal area, LNS)	NF	Myxoid stroma; fibroblastic cells (no atypia) + lymphocytes; infiltrative border	SMA S100-β (focal) Ki-67, 5%–10%	*MYH9*
2	M	30–35	Superficial (bilateral supraclavicular area)	NF	Oedematous collagen stroma; fibroblastic cells (no atypia) + lymphocytes; delineated border	SMA Ki-67, 5%	*MYH9*
3	F	25–30	Superficial (L inguinal area)	NF (IV)	Myxoid stroma; fibroblastic cells (no atypia) + osteoclast like cells; intravascular	SMA S100-β (focal) Ki-67, 1%	*MYH9*
4	F	45–50	Superficial (skin, LNS)	NF	Hyalinised stroma; fibroblastic cells (no atypia) + lymphocytes; delineated border	SMA Ki-67, 2%	*MYH9*
5	F	35–40	Deep (R zygomatic area)	NF	Myxoid stroma; fibroblastic cells (no atypia) + lymphocytes; infiltrative border	SMA S100-β (focal) Ki-67, 5%	*MYH9*
6	M	35–40	Superficial (R elbow)	NF	Myxoid to collagen stroma; fibroblastic cells (no atypia); delineated border	SMA Ki-67, 5%	*MYH9*
7	M	60–65	Superficial (R face)	NF	Myxoid to keloidal stroma; fibroblastic cells (no atypia); infiltrative border	SMA Ki-67, 5%	*MYH9*
8	M	50–55	Superficial (L supraclavicular area)	NF	Myxoid to collagen stroma; fibroblastic cells (no atypia) + lymphocytes; delineated border	SMA Ki-67, 3%	*MYH9*
9	F	15–20	Deep (R shoulder)	NF	Myxoid stroma; fibroblastic cells (no atypia); delineated border	SMA Ki-67, 2%	*MYH9*
10	F	30–35	Superficial (head, LNS)	NF (C)	Myxoid to keloidal stroma; fibroblastic cells (no atypia); infiltrative border	SMA Ki-67, 20%	*MYH9*
11	M	30–35	Superficial (L forearm)	NF	Myxoid to collagen stroma; fibroblastic cells (no atypia) + lymphocytes; infiltrative border	SMA Ki-67, 5%	*MYH9*
12	F	40–45	Superficial (L inguinal area)	NF	Myxoid to collagen stroma; fibroblastic cells (no atypia) + lymphocytes; delineated border	SMA Ki-67, 3%	*MYH9*
13	M	30–35	Superficial (hand, LNS)	NF	Myxoid to collagen stroma; fibroblastic cells (no atypia); delineated border	SMA Ki-67, 10%	*MYH9*

M, male; F, female; LNS, laterality not specified; L, left; R, right; NF, nodular fasciitis; IV, intravascular; C, cranial; IHC, immunohistochemistry; SMA, smooth muscle actin.

Table 2 Patient characteristics and clinical findings in cases with uncommon *USP6* fusion partners

14	F	25–30	Deep (tibial area, LNS)	NF	Myxoid to collagen stroma; fibroblastic cells (no atypia) + lymphocytes; infiltrative border	SMA Ki-67, 3%	*TMP4*
15	F	10–15	Superficial (R temporal area)	NF	Collagen stroma; fibroblastic cells (no atypia); infiltrative border	SMA Ki-67, 3%	*SPARC*
16	F	15–20	Superficial (L forearm)	NF	Myxoid stroma; fibroblastic cells (no atypia) + lymphocytes; infiltrative border	SMA Ki-67, 5%	*EIF5A*
17	M	10–15	Superficial (L arm)	NF	Keloidal stroma; fibroblastic cells (no atypia) + lymphocytes; infiltrative border	SMA Ki-67, 2%	*KIF1A*
18	M	0–5	Superficial (corner of the mouth, LNS)	NF (C)	Hyalinised stroma; fibroblastic cells (no atypia) + lymphocytes; infiltrative border	SMA Ki-67, 5%	*MIR22HG*
19	F	15–20	Superficial (R arm)	NF	Collagen stroma; fibroblastic cells (no atypia); delineated border	SMA Ki-67, 2%–3 %	*COL1A2*

M, male; F, female; LNS, laterality not specified; L, left; R, right; NF, nodular fasciitis; C, cranial; IHC, immunohistochemistry; SMA, smooth muscle actin.

localised superficially—specifically two of them in the supraclavicular region, two in the groin region, two in the forearm, two in the arm region and the rest evenly distributed by one among the following sites: temporal region, nuchal region, cheek, elbow, corner of the mouth, hand, head and skin otherwise non-specified (as it was sent for the second opinion as a consultation from another institution without further details regarding the locality of the lesion). Three of the cases (16%) were localised within deeper compartments—one close to the zygomatic bone, next one attached to the distal diaphysis of the tibia and the last one was localised within the muscle of the shoulder. For more details on patient characteristics, see tables 1 and 2.

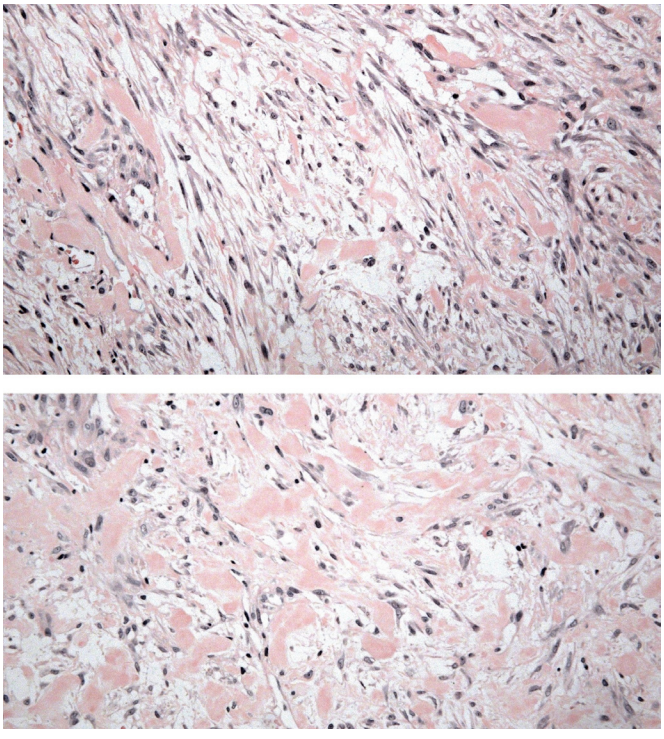

Figure 1 Light microscopy of a case of typical nodular fasciitis with keloidal change (H&E staining; ×200). This lesion consisted of plump spindle-shaped cells embedded in myxoid stroma with keloidal collagen bundles and showed typical tissue culture-like character. The neoplastic cells contain vesicular nuclei with visible nucleoli and lack hyperchromasia or pleomorphism.

Histological findings

All 19 cases met the morphological diagnostic criteria for a diagnosis of nodular fasciitis. Based on the localisation, three cases were further subclassified as cranial or intravascular subtypes of this lesion. All samples consisted of fascicular to storiform arranged plump spindle-shaped (myo)fibroblastic cells lacking nuclear hyperchromasia or pleomorphism. The cells contained plump spindle to oval-shaped nuclei with visible nucleoli. Mitotic figures were observed in each case, but none of them was atypical. The character of stroma varied—ranging from myxoid to collagenous with hyalinisation or even keloidal changes in three cases (16%) (figure 1). There was an admixture of lymphocytes within the interstitium in the majority of the samples (58%). One case of cranial fasciitis contained isolated osteoclast-like giant cells. Extravasation of erythrocytes was sometimes observed. The borders of the tumours were at least focally infiltrative in slight majority of the cases (53%), the rest of the lesions showed delineated borders with fibrotic capsule at the periphery. For more details on histopathological findings of all the cases, see tables 1 and 2.

Immunohistochemistry

In each of the 19 cases (100%), the tumourous spindle cells were SMA positive and negative for H-caldesmon and desmin (figure 2). Furthermore, three cases also showed weak and focal S100-β positivity. The proliferation index varied between 1% and 20% using the immunohistochemical marker Ki-67.

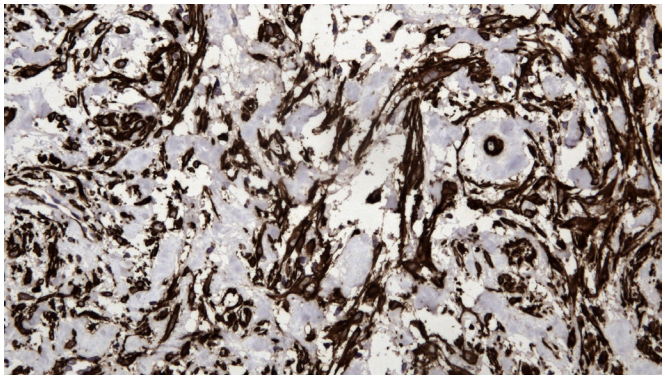

Figure 2 Light microscopy of a case of typical nodular fasciitis (SMA staining; ×200). All cases were positive in SMA using immunohistochemistry. SMA, smooth muscle-actin.

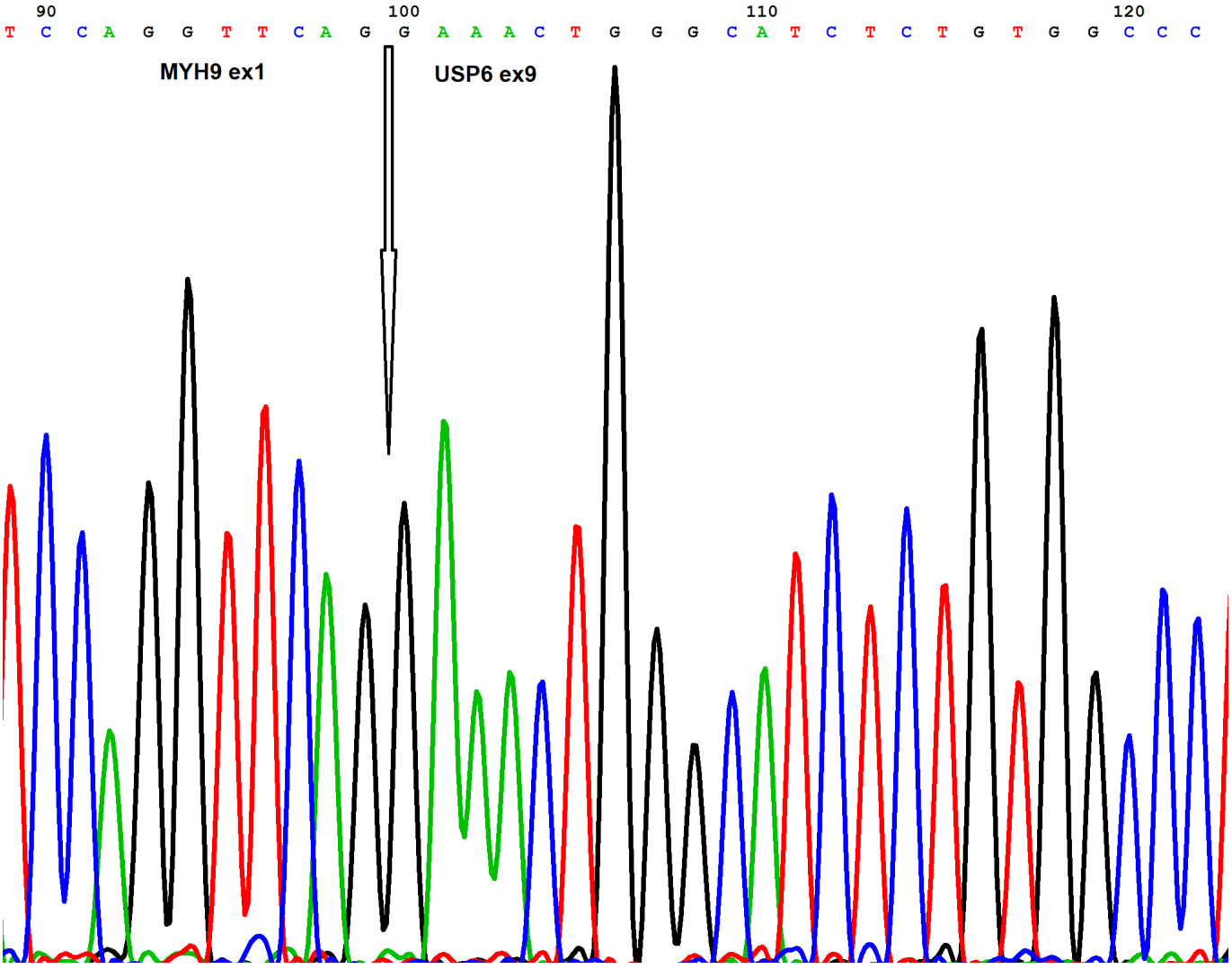

Figure 3 Sequencing analysis of *MYH9::USP6* positive nodular fasciitis.

Molecular findings

Molecular examination revealed the *USP6* rearrangement in all of the 19 cases (100%). Thirteen (68%) showed typical *MYH9::USP6* fusions (figure 3). Remaining six cases included *USP6* fusions with unusual partners (figure 4). The first one represented old female in late 20s with deep nodular fasciitis of the proximal tibial region in which *TPM4::USP6* fusion was proven. *EIF5A::USP6* fusion was found in the case of late adolescent female with superficial nodular fasciitis of the forearm region. Another unusual fusion—*SPARC::USP6* was detected in the case of girl in middle childhood with superficial nodular fasciitis of the temporal area. Next, in the case of an infant boy a superficial nodular fasciitis arose within mouth corner region harbouring a *MIR22HG::USP6* fusion. Next unusual fusion—*COL1A2::USP6* was found in the case of superficial nodular fasciitis of the arm region of early adolescent female. Eventually, the novel fusion *KIF1A::USP6* was identified in the case of early adolescent male with superficial nodular fasciitis of the arm region. This gene fusion in nodular fasciitis has not been reported in the literature to our knowledge so far. All of the described cases, including common and uncommon fusions, have shown benign clinical behaviour. During follow-up of the patients, there was only one episode of recurrence of the disease in case of female in her late 40s with nodular fasciitis of the skin, in which the typical *MYH9::USP6* fusion was proven (for more details, see table 1).

DISCUSSION

In 13 of 19 (68%) cases included in this study, the most common *MYH9::USP6* fusion was detected. Remaining 6 cases represented unusual *USP6* fusion partners such as *TPM4*, *EIF5A*, *SPARC*, *MIR22HG*, *COL1A2* and *KIF1A*. Five of these fusions have been already described in single case reports[9 13 14 17] and one represents a newly recognised fusion partner.

The molecular examination of nodular fasciitis has been proven as a useful tool aiming at correct diagnosis in selected cases,[3 6 7] as for nodular fasciitis can morphologically mimic a sarcomatous growth despite its benign nature. *USP6* is a gene located on chromosome 17p13 encoding a subfamily of deubiquitinating enzymes, the ubiquitin-specific proteases. They have various functions among human cells including intracellular turnover and intracellular trafficking.[18] *USP6* rearrangements with various fusion partners have been typically found in cases of nodular fasciitis, aneurysmal bone cysts and myositis ossificans.[16] For nodular fasciitis the typical fusion partner tends to be *MYH9*, which is a gene

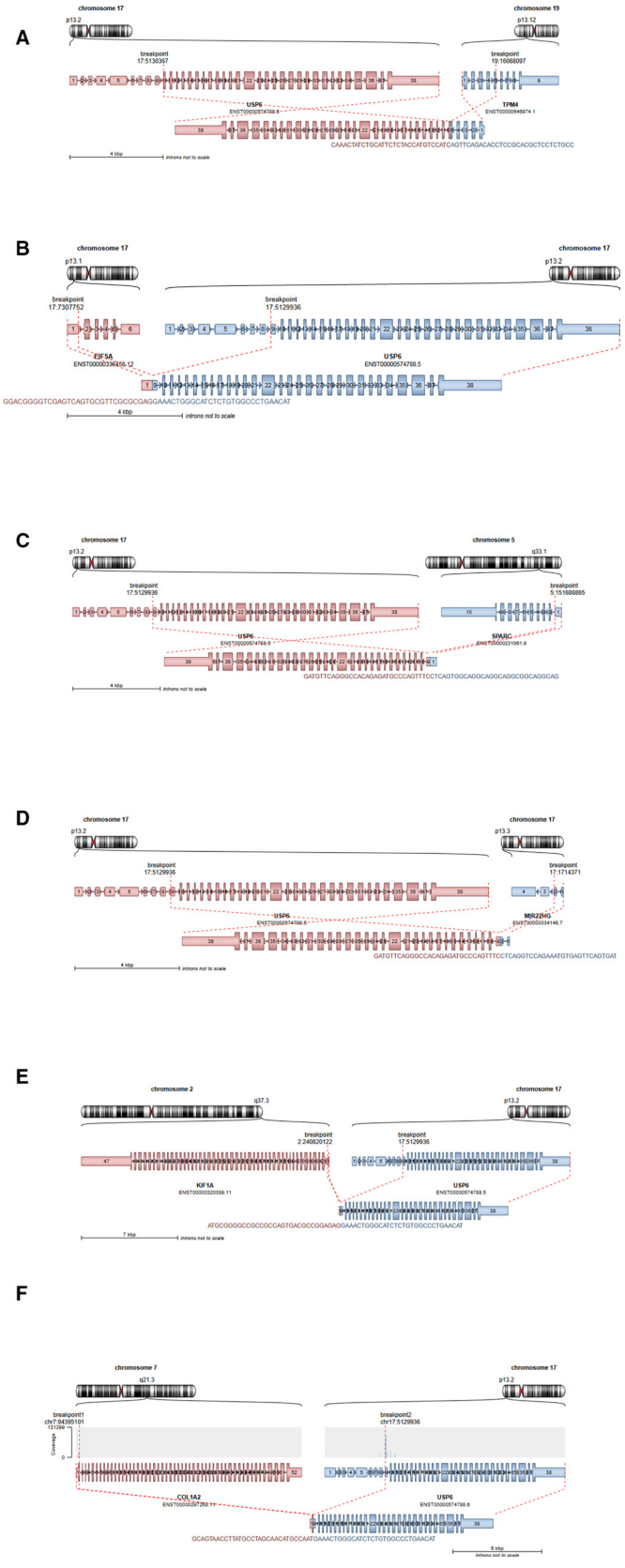

Figure 4 Schematic visualisation of detected fusion transcripts using Arriba software (https://github.com/suhrig/arriba/): (A) *TPM4::USP6* fusion partners; (B) *EIF5A::USP6* fusion partners; (C) *SPARC::USP6* fusion partners; (D) *MIR22::USP6* fusion partners; (E) *KIF1A::USP6* fusion partners; (F) *COL1A2::USP6* fusion partners.

encoding a non-muscle myosin IIA, an acting binding molecular motor helping to power the contraction of actin-myosin filaments.[26] Although *MYH9* expression has been found in many tumours it's exact role in neoplastic processes remains unclear.[26 27] Other described fusion partners of *USP6* are *TPM4*, *EIF5A*, *PPP6R3*, *CTNNB1*, *SPARC*, *THBS2*, *COL6A2*, *TNC*, *SEC31A*, *COL1A1*, *COL1A2*, *COL3A1*, *CALU*, *NACA*, *SLFN1*, *LDHA*, *SERPINH1*, *PDLIM7*, *MYL12A*, *PAFAH1B1* and *MIR22HG* as previously mentioned.[9 12–25] In this rapidly changing landscape of new molecular findings, there has been a growing amount of cases describing morphologically and even clinically malignant cases of nodular fasciitis correlating with different *USP6* fusion partners.[12 19 23] In this study, five rare (*TPM4*, *SPARC*, *EIF5A*, *COL1A2* and *MIR22HG*) and one novel (*KIF1A*) fusion partners have been identified and in each of these cases, nodular fasciitis showed benign clinical behaviour. During follow-up of all the patients, there was only one episode of recurrence of the disease in case of female in late 40s with nodular fasciitis of the skin, in which the typical *MYH9::USP6* fusion was proven (for more details, see table 1).

EIF5A is a gene that encodes translation initiation factor 5A-1 involved in maintenance of cell wall integrity, apoptosis and other functions. Previously described case involves a female in her 40s with a forearm subcutaneous mass.[14] Our case represented late adolescent female with a superficial subcutaneous mass also in forearm region.

TPM4 is an actin binding protein participating in muscle contraction. Previously described case involved a male in early 30s with a cheek mass.[13] Our case included a female in late 20s with deep nodular fasciitis within the proximal tibial region.

MIR22HG is a gene connected closely to miRNA class, involved in post-transcriptional regulation. Previously described case presented male in late 30s with subscapular mass.[9] Our case represented an infant boy with a mass localised in the mouth corner region.

SPARC is a gene that encodes cysteine-rich acidic matrix-associated protein that plays an important role in collagen calcification within bones and also other processes within extracellular matrix. Previously described case involved a male in late 50s with a mass affecting tendons of the third and fourth finger of his left hand.[9] Our case represents a girl in middle childhood with superficially growing nodular fasciitis of the temporal region.

COL1A2 encodes one chain of collagen type I and is commonly present in vast majority of human connective tissue and is also expressed in some human tumours.[28] Previously described case presented a cervical nodular fasciitis of a girl around age 1.[17] Our case represents early adolescent female with superficially growing nodular fasciitis of the arm region.

KIF1A is a member of 1A kinesin family and it encodes a protein participating in axonal transport. It has been described that its mutations play a major role in hereditary sensory neuropathy IIC and spastic paraplegia 30 and are also associated with a development of amyotrophic lateral sclerosis.[29] Case of *KIF1A* fusion with *USP6* in terms of nodular fasciitis has never been described before. We report *KIF1A::USP6* fusion in the case of early adolescent boy with superficially growing nodular fasciitis of the arm.

In summary, this study presents 19 cases of morphologically, immunohistochemically and molecularly confirmed cases of nodular fasciitis including five rare and one novel *USP6* fusion partners, which can help to understand the process of development of such lesion. Moreover, it can be useful in routine

histopathological practice of soft-tissues diagnostics, preventing possible misdiagnosis of malignancy.

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Contributors JB, LK and JZ designed the report. JB and MS analysed the data, and wrote major part of the manuscript. LK and JZ contributed to the writing of the manuscript. JB performed the pathological diagnosis, immunohistochemistry and prepared histopathological figures. LK performed molecular analysis. JZ is the guarantor. All authors edited the final manuscript. All authors read and approved the final manuscript.

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Competing interests None declared.

Patient consent for publication Not applicable.

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