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# A patient with tumor necrosis factor receptor-associated periodic syndrome misdiagnosed as Kawasaki disease: A case report and literature review

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ABSTRACT

This article reports a case of tumor necrosis factor receptor-associated periodic syndrome (TRAPS) misdiagnosed as Kawasaki disease and summarizes the clinical features and therapeutic progress of TRAPS and the relationship between its clinical manifestations and gene mutations. We retrospectively analyzed a patient with tumor necrosis factor receptor superfamily member 1A (TNFRSF1A) -mutated auto-inflammatory disease who was misdiagnosed with Kawasaki disease in another hospital. The clinical features and therapeutic progress of TRAPS were analyzed by combining clinical features and gene reports of this case and literature review. TRAPS onset occurred in a female pediatric patient at the age of 4 months. The child and in his father at the age of 6 years, both of whom manifested periodic fever, and recurrent rash, as well as elevated leukocytes, erythrocyte sedimentation rate (ESR), and C-reactive protein (CRP) during episodes but normal between episodes. This child carried a heterozygous mutation in TNFRSF1A located in the region 6442923–6442931 on chromosome 12. The nucleic acid alteration was: c.298 (exon3) _c.306 (exon3) 291 delCTCAGCTGC, resulting in a 3 amino acid deletion p.L100_C 102del 292 (p.Leu100_Cys102del) (NM_001065). After etanercept treatment, the symptoms of fever and rash disappeared, and the levels of ESR, CRP, interleukin (IL)-1, IL-6, and TNF-α levels were normal. Subsequently, no liver, kidney, or cardiac amyloidosis and severe etanercept-related adverse events were observed at 1-year follow-up. TRAPS pathogenesis is associated with TNFRSF1A mutation, which is characterized by periodic episodes of fever, mostly accompanied by recurrent rashes, periorbital edema, abdominal pain, and serious complications of organ amyloidosis. Moreover, etanercept can effectively alleviate the clinical symptoms and high inflammation level of TRAPS.

## 1. Introduction

Tumor necrosis factor receptor-associated periodic syndrome (TRAPS) is a rare autosomal dominant auto-inflammatory disease caused by a heterozygous mutant of tumor necrosis factor (TNF) receptor superfamily member 1A (TNFRSF1A) on chromosome 12,

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which is characterized by periodic episodes of fever associated with additional symptoms, including rash, arthralgia, periorbital edema, lymphadenectasis, and hepatosplenomegaly. In TRAPS, acute phase reactants are markedly elevated during recurrent or persistent episodes of inflammation but completely normal between episodes with complete disappearance of symptoms. TRAPS is distinguished from diffuse connective tissue diseases by the absence of high titer autoantibodies or abnormal activation of specific T cells. Accordingly, this disease lacks specific clinical manifestations and is prone to misdiagnosis and underdiagnosis. Most TRAPS patients have a favorable prognosis, while severe cases can develop amyloidosis, causing a poorer prognosis. This article reported the clinical data of a child with TRAPS admitted in 2022 who was initially misdiagnosed with Kawasaki disease and summarized the difference between the clinical features of this disease and Kawasaki disease to improve the understanding of this disease in clinical practice.

## 2. Case presentation

A female child aged 1 year and 1 month old was admitted to our department on December 4, 2021 with "recurrent fever with rash for 10 months and recurrence for 1 week". The child developed an unexplained fever 10 months ago, which peaked at 40 °C 2–3 times per day and could be reduced to normal temperature after treatment with non-steroidal anti-inflammatory drugs (NSAIDs). The fever was often accompanied by chills and rashes which mostly occurred after about 5–7 days of fever, appeared on the chest, abdomen, and posterior back as maculopapular rashes that faded under pressure and were not itchy and were significantly relieved when the fever subsided. This child had no significant cough, expectoration, abdominal pain, diarrhea, headache, dizziness, recurrent aphthous ulcer, myalgia, painful swelling of joints, or periorbital edema. He visited the local hospital several times, showed elevated leukocytes and C-reactive protein (CRP) in routine blood tests, and was discharged with a normal temperature after anti-infection treatment(administered sequentially with Ceftizoxime and cefoperazone and sulbactam for antimicrobial treatment). However, fever and rashes recurred shortly after discharge. Three months ago, the patient recured with fever and rashes and consulted a people's hospital in our city, where anti-infection treatment was given and ineffective and Kawasaki disease was considered. Therefore, he was given 2nd gamma globulin pulse therapy and aspirin treatment and discharged with a normal temperature. Nevertheless, this child suffered from a recurrence of fever and rashes 1 week ago and was admitted to our department. The child had no seasonal or diurnal variations in fever, no abnormal birth history, and the same intellectual and growth development as normal children of the same age. His family history was as follows: his father visited another hospital at the age of 6 years for "recurrent fever, rashes, and arthritis", was diagnosed with "still's disease", and received effective hormone treatment, but recurrent episodes have been occurring since hormones (prednisone, at a dose of 15mg/day) were discontinued.

Physical examination on admission revealed a temperature of 38.2 °C, a pulse rate of 120 times/min, a height of 76 cm, a weight of 11 kg, clear consciousness, responsiveness, maculopapular rash scattered on the trunk and extremities that faded under pressure. In addition, several bean-like lymph nodes were palpable in the bilateral neck, which were hard in texture and movable without tenderness. The heart, lungs, abdomen, and neurological examination exhibited no abnormality. There was no painful swelling of joints in the extremities.

The following auxiliary tests were conducted. First, the results of laboratory tests were listed below (Table 1): the blood routine test showed 22.35 $\times$ $10^9$/L white blood cells (WBC), 15.15 $\times$ $10^9$/L neutrophil, 106 g/L hemoglobin, 609 $\times$ $10^9$/L platelets, 36 mm/h erythrocyte sedimentation rate (ESR), 109.7 mg/L CRP, and 544.7 mg/L serum amyloid A; the cytokine test demonstrated 7.7 pg/mL interleukin (IL) -β, 1993 U/mL IL-2 receptor, 14 pg/mL IL-10, and 25 pg/mL IL-6; the test of lymphocyte subsets of revealed a percentage of $CD3^+$ T lymphocytes of 55.9%, a percentage of $CD3^+CD8^+$ T lymphocytes of 13.8%, a ratio of $CD3^+CD4^+$ T lymphocytes/$CD3{+}CD8^+$ T lymphocytes of 2.73, a percentage of $CD3^-CD16{+}CD56^+$ NK cells of 6.9%, a percentage of $CD3^+CD19^+$ B cells of 35.9%, an absolute count of $CD3^+$ T lymphocytes of 4280 cells/μL, an absolute count of $CD3^+CD4^+$ T lymphocytes of 2888 cells/μL, an absolute count of $CD3^+CD8{+}$T lymphocytes of 1059 cells/μL, an absolute count of $CD3^-CD19^+$ B cells of 2747 cells/μL; the concentration of complement C3 and C4 was 2.07 g/L and 0.35 g/L, respectively; no abnormality was observed in the tests of biochemistry, cardiac enzymes, immunoglobulins, rheumatoid factor, synovial fluids, five blood coagulation indexes, antinuclear antibody, antineutrophil autoantibody, and five tumor markers, as well as in routine urine and stool tests. Second, etiological examinations, including Epstein-Barr (EB)-DNA, EB virus antibody, cytomegalovirus (CMV)-DNA, CMV antibody, human immunodeficiency virus, hepatitis B, hepatitis C, purified protein derivative, T-spot, blood culture, and bone marrow culture tests, showed no abnormalities. Third, the results of ultrasound and imaging, including abdominal ultrasound, cardiac ultrasound, cranial magnetic

**Table 1**
The laboratory indicators for pediatric patients.

| laboratory indicators | Reference range | Time | | | | |
|---|---|---|---|---|---|---|
| | | Before treatment | 1 month after treatment | 3 months after treatment | 6 months after treatment | 1 year after treatment |
| WBC ($10^9$/L) | 4.6–12.7 | 22.35 | 15.6 | 12.5 | 10.3 | 8.4 |
| ESR (mm/h) | 0–20 | 36 | 20 | 16 | 15 | 8 |
| CRP (mg/L) | 0–8 | 109.7 | 25 | 10.4 | 7.8 | <0.5 |
| PLT ($10^9$/L) | 0–300 | 609 | 486 | 342 | 298 | 268 |
| SAA ( mg/L ) | 0–10 | 544.7 | 68.4 | 25.9 | 3.3 | 3.1 |

Abbreviation: WBC, white blood cells; ESR, erythrocyte sedimentation rate; CRP, C-reactive protein; PLT, platelet; SAA, serum amyloid A.

resonance imaging, and chest computed tomography, were all normal. Fourth, the genetic examination was performed: after informed consent was obtained from the parents of the child, 3 mL peripheral blood was taken from the child and his parents for whole-exome genetic testing, which indicated that the child carried a heterozygous mutation in TNFRSF1A located in the region 6442923–6442931 on the chromosome 12, his father carried a heterozygous mutation, and his mother was wild type. The alteration in the nucleic acid sequence is as follows: c.298 (exon3) _c.306 (exon3), resulting in a deletion of three amino acids. This includes p.L100_C102del292 (p. Leu100_Cys102del) (NM_001065). (Fig. 1*A.,B,C*).

The treatment processes were as follows: based on the medical history and genetic test results, the child was clearly diagnosed with TRAPS and treated with oral ibuprofen combined with the TNF antagonist etanercept (12.5 mg/10 days, subcutaneous injection, 0.8 mg/kg per week). After treatment, the body temperature returned to normal and the patient was discharged. No abnormalities were found in routine blood, ESR, CRP, liver and kidney function, and cytokine tests during 1 year of regular outpatient follow-up. Before treatment, the Activity Index of Periodic Fever Syndrome (AIDAI) score for the first month was 28 points (fever: 14 points + rash: 14 points). After initiating treatment, the AIDAI score decreased to 4 points in the first month and eventually reached 0 points in the third month.

## 3. Discussion

The case of TRAPS was reported first in Ireland in 1982 [1] and then all over the world, mostly in Europe and relatively few in Asia. We reported a case of TRAPS misdiagnosed as Kawasaki disease at the early clinical stage and summarize the pathogenesis of the disease, the characteristics of its clinical manifestations, and the progress of its diagnosis and management by combining with literature review, thus improving the understanding of pediatricians for the disease, reducing the misdiagnosis rate, and preventing serious complications such as amyloidosis.

Initially, the child in this case report had a short fever episode before being admitted to the hospital. Upon admission, they exhibited symptoms such as fever, rash, elevated WBC count, high levels of CRP and ESR. Despite receiving anti-infection treatment, it proved to be ineffective. However, two doses of gamma globulin pulse therapy and aspirin anti-inflammatory therapy were found to be effective. Additionally, a detailed family history was not obtained during the assessment process. Therefore, based on the exclusion of common infectious diseases and clinical evaluation, the patient was clinically diagnosed with Kawasaki disease. It is important to note that the clinical manifestations of Kawasaki disease include rash, bulbar conjunctival congestion, strawberry tongue, swelling of the skin and mucosa in the hands and feet. However, this pediatric patient exhibited a prolonged, recurrent, periodic fever and presented

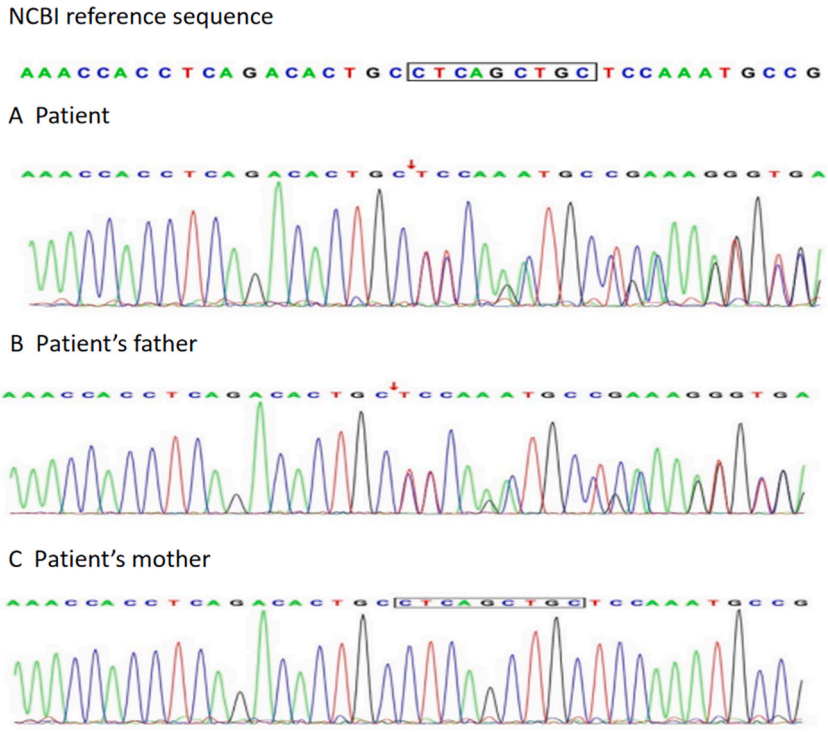


**Fig. 1.** The sanger Sequencing chromatagram of the variants. According to the Human Genome Variation Society (HGVS) rule, this mutation was validated by Sanger sequencing, referring to the chromosome version (hg19). The mutation in the affected child (A)is a heterozygous mutation in the TNFRSF1A gene located in the region 6442923–6442931 on chromosome 12, with a heterozygous mutation in the father (B)and wild type in the mother ( C ) . The nucleic acid alteration was as follows: c.298 (exon3) _c.306 (exon3) delCTCAGCTGC, and the amino acid alteration was as follows: p.L100_C 102del (p.Leu100_Cys102del) (NM_001065).

solely with a rash, accompanied by atypical coronary dilation of the heart. Following treatment with C-ball and aspirin, their body temperature temporarily returned to normal. However, the fever soon reappeared. Furthermore, the patient's father experienced similar clinical manifestations during his youth. Based on the clinical characteristics of this case, genetic reports, and a literature review, the final diagnosis was determined to be TRAPS.

TRAPS is a rare autosomal dominant auto-inflammatory disease, with unclear and partially controversial specific pathogenetic mechanisms. The causative gene for this disease was revealed to be located on chromosome 12p13 and then confirmed in further studies as TNFRSF1A [2,3]. To date, 43 missense mutations in TNFRSF1A have been validated as pathogenic, with 56 probable pathogenic mutations, in the Infevers database [4]. The retention of TRAPS mutant TNFR1 molecules intracellularly and colocalisation with endoplasmic reticulum (ER) markers was first reported by Lobito AA et al. [5]. In addition, almost all known TRAPS-associated mutations are point mutations, which result in amino acid substitutions in the extracellular structural domain of TNFR1 [6]. Although some TRAPS-associated mutations, such as T79 M and C62Y, have been identified as pathogenic, mechanisms underlying recurrent episodes of inflammation remain unclear. Of note, a study [7] has demonstrated that due to mutations in TNFRSF1A, the TNF receptor fails to reach the cell surface but can be trapped by the endoplasmic reticulum, which causes inflammation in vivo and increases the expression of pro-inflammatory cytokines. In addition, another study elucidated that misfolded TNFR1 was retained within the endoplasmic reticulum, contributing to a decrease in TNF-induced apoptosis, an increase in intrinsic immune stimulation, and then an elevation in inflammatory cytokines, such as TNF-α, IL-6, and IL-1β [8]. However, a study of a Japanese scholar [9] on TNFRSF1A mutant (T79 M, G87V, or T90I) mice revealed that T79 M and G87V mutations attenuated the inflammatory response to TNF-α and that TNFR1 levels were increased in whole-cell lysates of TRAPS mutant macrophages but markedly reduced in the surface of TRAPS mutant macrophages. This result suggested that TRAPS mutations did not enhance the inflammatory response to TNF-α and lipopolysaccharide but, instead, suppressed the response to TNF-α by diminishing the cell surface expression of TNFR1. Accordingly, it could be speculated that TRAPS-associated inflammation may be induced by unconfirmed disease-specific pro-inflammatory factors. In summary, the specific pathogenetic mechanism underlying TRAPS is unknown and needs to be further investigated.

Moreover, some papers have reported deletions/duplications of the TNFRSF1A gene in relation to TRAPS [10–13]. A study identified twenty-one patients with a variant of the TNFRSF1A gene. Among these patients, thirteen exhibited an R92Q substitution, while eight had other missense substitutions. Furthermore, one patient presented a splicing mutation, and another patient had an in-frame interstitial deletion [10]. Churchman SM et al. have discovered a novel mutation, c.472+1G > A (C158delinsYERSSPEAKPSPHPRG), which affects a splice site in intron 4 of the TNFRSF1A gene. This mutation leads to a 45-nucleotide insertion of intronic DNA into the mRNA [11]. Of the 30 patients diagnosed with idiopathic recurrent pericarditis and exhibiting a poor response to colchicine treatment, four individuals were found to have TNFRSF1A mutations. Among these four patients, one presented a novel heterozygous deletion (DeltaY103-R104), while the remaining three patients carried a heterozygous low-penetrance R92Q mutation [12]. In a Brief Report, an association was discovered between TRAPS and gonosomal mosaicism involving a novel 24-nucleotide deletion in the TNFRSF1A gene. The presence of this deletion (c.255_278del) in the TNFRSF1A gene was confirmed through Sanger sequencing, which was further supported by allele-specific PCR [13].

As reported, TRAPS mostly develops before the age of 10 years (a median onset age of 4.3 years) and occurred in 10% of patients in adulthood with mild symptoms [7,14]. The study conducted by Zhao M et al. included nine adult patients (aged ≥16 years) with TRAPS. It reported that the median age of disease onset was 3 years (range: 0.5–38.5), and adult-onset TRAPS was observed in two (22.2%) patients. The median time of diagnosis delay was 16.5 years (range: 1.5–50.5). Nine TNFRSF1A gene variants were detected, namely C58R, G65E, F89L, C99G, V202G, V202D, c.769-23T > C, S290I, and c.*64T > C. Rash was more frequently observed in Chinese patients compared to Japanese and European patients, whereas chest pain and amyloidosis occurred less frequently [15]. Additionally, Yichao Hua et al. have found 5 patients with TRAPS among 92 Chinese adult patients with systemic autoinflammatory diseases [16].

TRAPS is typically characterized clinically by recurrent episodes of fever and inflammation in different parts of the body, primarily manifesting as fever, myalgia, erythema migrans, periorbital edema, and abdominal pain, and its prognosis depends on amyloidosis secondary to recurrent episodes of inflammation [6]. In TRAPS patients, febrile episodes occur every 4–6 weeks and last 5 days–3 weeks, which may be associated with emotions, stress, infection, and trauma, with most triggers still unknown [17–20]. During the episode, 75% of patients may present with cutaneous symptoms which manifest as erythema migrans, urticaria, and erysipelas-like rashes and are histologically characterized by a dermal infiltrate of lymphocytes and monocytes, and immunohistochemistry reveals that the infiltrate consists of T cells ($CD3^+$, $CD4^+$, $CD8^+$) and monocytes ($CD68^+$) and is negative for B cells and multinucleated macrophages [21]. TRAPS often involves the eye (major manifestations include conjunctivitis and unilateral or bilateral periorbital pain and edema), gastrointestinal tract (the major manifestation is abdominal pain secondary to inflammatory peritonitis), and joints (arthralgia is the major manifestation and arthritis is rare) [22]. The most severe complication of TRAPS is amyloidosis secondary to recurrent episodes of inflammation over several years, and TRAPS patients with cysteine mutations are at higher risk of developing amyloidosis than those carrying non-cysteine mutations [23]. In our report, this case clinically manifested as recurrent and periodic episodes of fever and maculopapular rashes, without ocular and gastrointestinal involvement and AA amyloidosis, suggesting to some extent that there is a large degree of heterogeneity among TRAPS patients with different gene mutations and those with the same mutation [23].

Early identification and diagnosis of TRAPS are ultimately aimed at providing patients with timely treatment to control symptoms, relieve episodes, and prevent serious complications including amyloidosis. At present, TRAPS lacks standardized and unified treatment regimens. In 2022, the Rheumatology Group, Pediatrics Branch of the Chinese Medical Association put forward an expert consensus on the diagnosis and treatment of auto-inflammatory diseases in children. Currently, the main treatment for acquired immunodeficiency syndrome is traditional treatment and biologic targeted therapy [24]. Reportedly, NSAIDs combined with high doses of corticosteroids

can effectively reduce inflammation in the short term, whereas long-term use of corticosteroids fails to effectively reduce the frequency or intensity of episodes of inflammation and cannot prevent the occurrence of amyloidosis [22]. Moreover, etanercept can effectively repress inflammation, decrease the frequency and duration of episodes of inflammation and the dosage of corticosteroids, and effectively prevent AA amyloidosis in some patients. Nevertheless, the efficacy of etanercept varies among patients [25,26]. The poor clinical response of TRAPS patients to anti-TNF monoclonal antibodies may be related to the occurrence of a paradoxical inflammatory response [27,28]. A multicenter retrospective study in Italy unraveled that the majority of TRPAS patients, even some patients with etanercept treatment failure, effectively responded to IL-1 inhibitors such as anakinra and canakinumab, without serious adverse events at long-term follow-up [29]. Furthermore, other studies elaborated that the IL-6 inhibitor tocilizumab could be utilized for the treatment of TRAPS patients who failed to respond to TNF and IL-1 inhibitors [30, 31]. Nonetheless, no studies have been conducted to explore how to prolong the episode cycle and improve the prognosis for TRAPS.

## 4. Conclusion

TRAPS is one of the auto-inflammatory diseases caused by mutations in single genes. Despite significant progress in the study of TRAPS pathogenesis at the genetic level, the specific pathogenetic mechanism needs to be further elucidated and there is no expert consensus and guidelines for the diagnosis and treatment of this disease at home and abroad. In clinical practice, TRAPS is usually misdiagnosed as sepsis, Kawasaki disease, and systemic juvenile idiopathic arthritis due to clinical symptoms such as fever and rashes and non-specific indicators of high-level inflammation, and long-term chronic inflammation can cause serious complications including amyloidosis. Through the combination of this case and literature review, we can understand the clinical features and treatment progress of TRAPS, which improves the understanding of clinical pediatricians for the disease, reduces misdiagnosis, and prevents the occurrence of severe complications.

## Ethics statement

This study was designed that followed were in accordance with the ethical standards of the responsible committee on human experimentation (Anhui Provincial Children's Hospital) on December 18, 2018(APPROVAL NUMBER: EYLL-2018-20) and with the Helsinki Declaration of 1964, as revised in 2013. Written consent had obtained from parents or legal guardians for publication.

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## Author contribution statement

All authors listed have significantly contributed to the investigation, development and writing of this article.

## Data availability statement

Data will be made available on request.

## Declaration of competing interest

The authors declare that they have no known competing financial interests or personal relationships that could have appeared to influence the work reported in this paper.

## References

[1] L.M. Williamson, D. Hull, R. Mehta, W.G. Reeves, B.H. Robinson, P.J. Toghill, Familial Hibernian fever, Q. J. Med. 51 (1982) 469–480.
[2] M.F. McDermott, B.W. Ogunkolade, E.M. McDermott, L.C. Jones, Y. Wan, K.A. Quane, et al., Linkage of familial Hibernian fever to chromosome 12p13, Am. J. Hum. Genet. 62 (1998) 1446–1451, https://doi.org/10.1086/301886.
[3] M.F. McDermott, I. Aksentijevich, J. Galon, E.M. McDermott, B.W. Ogunkolade, M. Centola, et al., Germline mutations in the extracellular domains of the 55 kDa TNF receptor, TNFR1, define a family of dominantly inherited autoinflammatory syndromes, Cell 97 (1999) 133–144, https://doi.org/10.1016/s0092-8674(00)80721-7.
[4] M.E. Van Gijn, I. Ceccherini, Y. Shinar, E.C. Carbo, M. Slofstra, J.I. Arostegui, et al., New workflow for classification of genetic variants' pathogenicity applied to hereditary recurrent fevers by the International Study Group for Systemic Autoinflammatory Diseases (INSAID), J. Med. Genet. 55 (2018) 530–537, https://doi.org/10.1136/jmedgenet-2017-105216.
[5] A.A. Lobito, F.C. Kimberley, J.R. Muppidi, H. Komarow, A.J. Jackson, K.M. Hull, et al., Abnormal disulfide-linked oligomerization results in ER retention and altered signaling by TNFR1 mutants in TNFR1-associated periodic fever syndrome (TRAPS), Blood 108 (15) (2006) 1320–1327, https://doi.org/10.1182/blood-2005-11-006783.
[6] C. Cudrici, N. Deuitch, I. Aksentijevich, Revisiting TNF receptor-associated periodic syndrome (TRAPS): current perspectives, Int. J. Mol. Sci. 21 (2020), https://doi.org/10.3390/ijms21093263.
[7] T. Pettersson, J. Kantonen, S. Matikainen, H. Repo, Setting up TRAPS, Ann. Med. 44 (2012) 109–118, https://doi.org/10.3109/07853890.2010.548399.
[8] D. Rigante, G. Lopalco, A. Vitale, O.M. Lucherini, C. De Clemente, F. Caso, et al., Key facts and hot spots on tumor necrosis factor receptor-associated periodic syndrome, Clin. Rheumatol. 33 (2014) 1197–1207, https://doi.org/10.1007/s10067-014-2722-z.

[9] T. Akagi, S. Hiramatsu-Asano, K. Ikeda, H. Hirano, S. Tsuji, A. Yahagi, et al., TRAPS mutations in Tnfrsf1a decrease the responsiveness to TNFalpha via reduced cell surface expression of TNFR1, Front. Immunol. 13 (2022), 926175, https://doi.org/10.3389/fimmu.2022.926175.
[10] A. D'Osualdo, F. Ferlito, I. Prigione, L. Obici, A. Meini, F. Zulian, et al., Neutrophils from patients with TNFRSF1A mutations display resistance to tumor necrosis factor-induced apoptosis: pathogenetic and clinical implications, Arthritis Rheum. 54 (2006) 998–1008, https://doi.org/10.1002/art.21657.
[11] S.M. Churchman, L.D. Church, S. Savic, L.R. Coulthard, B. Hayward, B. Nedjai, et al., A novel TNFRSF1A splice mutation associated with increased nuclear factor kappaB (NF-kappaB) transcription factor activation in patients with tumour necrosis factor receptor associated periodic syndrome (TRAPS), Ann. Rheum. Dis. 67 (2008) 1589–1595, https://doi.org/10.1136/ard.2007.078667.
[12] L. Cantarini, O.M. Lucherini, R. Cimaz, C.T. Baldari, F. Bellisai, S. Rossi Paccani, et al., Idiopathic recurrent pericarditis refractory to colchicine treatment can reveal tumor necrosis factor receptor-associated periodic syndrome, Int. J. Immunopathol. Pharmacol. 22 (2009) 1051–1058, https://doi.org/10.1177/039463200902200421.
[13] D.M. Rowczenio, H. Trojer, E. Omoyinmi, J.I. Aróstegui, G. Arakelov, et al., Brief report: association of tumor necrosis factor receptor-associated periodic syndrome with gonosomal mosaicism of a novel 24-nucleotide TNFRSF1A deletion, Arthritis Rheumatol. 68 (2016) 2044–2049, https://doi.org/10.1002/art.39683.
[14] H.J. Lachmann, Periodic fever syndromes, Best Pract. Res. Clin. Rheumatol. 31 (2017) 596–609, https://doi.org/10.1016/j.berh.2017.12.001.
[15] M. Zhao, Y. Luo, D. Wu, Y. Yang, Y. Sun, R. Wang, et al., Clinical and genetic features of Chinese adult patients with tumour necrosis factor receptor-associated periodic fever syndrome, Rheumatology 59 (2020) 1969–1974, https://doi.org/10.1093/rheumatology/kez569.
[16] Y. Hua, D. Wu, M. Shen, K. Yu, W. Zhang, X. Zeng, Phenotypes and genotypes of Chinese adult patients with systemic autoinflammatory diseases, Semin. Arthritis Rheum. 49 (2019) 446–452, https://doi.org/10.1016/j.semarthrit.2019.05.002.
[17] H.J. Lachmann, R. Papa, K. Gerhold, L. Obici, I. Touitou, L. Cantarini, et al., The phenotype of TNF receptor-associated autoinflammatory syndrome (TRAPS) at presentation: a series of 158 cases from the Eurofever/EUROTRAPS international registry, Ann. Rheum. Dis. 73 (2014) 2160–2167, https://doi.org/10.1136/annrheumdis-2013-204184.
[18] E. Ruiz-Ortiz, E. Iglesias, A. Soriano, S. Bujan-Rivas, M. Espanol-Rego, R. Castellanos-Moreira, et al., Disease phenotype and outcome depending on the age at disease onset in patients carrying the R92Q low-penetrance variant in TNFRSF1A gene, Front. Immunol. 8 (2017) 299, https://doi.org/10.3389/fimmu.2017.00299.
[19] A. Nezos, O.D. Argyropoulou, E. Klinaki, N. Marketos, P. Karagianni, E. Eliopoulos, et al., Molecular and clinical spectrum of four pedigrees of TRAPS in Greece: results from a national referral center, Rheumatology 59 (2020) 1241–1246, https://doi.org/10.1093/rheumatology/kez424.
[20] M. Zhao, Y. Luo, D. Wu, Y. Yang, Y. Sun, R. Wang, et al., Clinical and genetic features of Chinese adult patients with tumour necrosis factor receptor-associated periodic fever syndrome, Rheumatology 59 (2020) 1969–1974, https://doi.org/10.1093/rheumatology/kez569.
[21] R. Schmaltz, T. Vogt, J. Reichrath, Skin manifestations in tumor necrosis factor receptor-associated periodic syndrome (TRAPS), Dermatoendocrinol 2 (2010) 26–29, https://doi.org/10.4161/derm.2.1.12387.
[22] K.M. Hull, E. Drewe, I. Aksentijevich, H.K. Singh, K. Wong, E.M. McDermott, et al., The TNF receptor-associated periodic syndrome (TRAPS): emerging concepts of an autoinflammatory disorder, Medicine (Baltim.) 81 (2002) 349–368, https://doi.org/10.1097/00005792-200209000-00002.
[23] S. Savic, L.J. Dickie, M. Battellino, M.F. McDermott, Familial Mediterranean fever and related periodic fever syndromes/autoinflammatory diseases, Curr. Opin. Rheumatol. 24 (2012) 103–112, https://doi.org/10.1097/BOR.0b013e32834dd2d5.
[24] The Rheumatology Group, Pediatrics Branch of the Chinese Medical Association. Exerts consensus on diagnosis and treatment of autoinflammatory diseases in children, Chinese J. of Appl. Clin. Pediatr. 37 (2022) 161–172.
[25] A.C. Bulua, D.B. Mogul, I. Aksentijevich, H. Singh, D.Y. He, L.R. Muenz, et al., Efficacy of etanercept in the tumor necrosis factor receptor-associated periodic syndrome: a prospective, open-label, dose-escalation study, Arthritis Rheum. 64 (2012) 908–913, https://doi.org/10.1002/art.33416.
[26] S. Stojanov, C. Dejaco, P. Lohse, K. Huss, C. Duftner, B.H. Belohradsky, et al., Clinical and functional characterisation of a novel TNFRSF1A c.605T>A/V173D cleavage site mutation associated with tumour necrosis factor receptor-associated periodic fever syndrome (TRAPS), cardiovascular complications and excellent response to etanercept treatment, Ann. Rheum. Dis. 67 (2008) 1292–1298, https://doi.org/10.1136/ard.2007.079376.
[27] B. Nedjai, G.A. Hitman, N. Quillinan, R.J. Coughlan, L. Church, M.F. McDermott, et al., Proinflammatory action of the antiinflammatory drug infliximab in tumor necrosis factor receptor-associated periodic syndrome, Arthritis Rheum. 60 (2009) 619–625, https://doi.org/10.1002/art.24294.
[28] Z. Kaymakcalan, P. Sakorafas, S. Bose, S. Scesney, L. Xiong, D.K. Hanzatian, et al., Comparisons of affinities, avidities, and complement activation of adalimumab, infliximab, and etanercept in binding to soluble and membrane tumor necrosis factor, Clin. Immunol. 131 (2009) 308–316, https://doi.org/10.1016/j.clim.2009.01.002.
[29] A. Vitale, L. Obici, M. Cattalini, G. Lopalco, G. Merlini, N. Ricco, et al., Biotechnological agents for patients with tumor necrosis factor receptor associated periodic syndrome-therapeutic outcome and predictors of response: real-life data from the AIDA network, Front. Med. 8 (2021), 668173, https://doi.org/10.3389/fmed.2021.668173.
[30] M.S. Akasbi N Soyfoo, Successful treatment of tumor necrosis factor receptor-associated periodic syndrome (TRAPS) with tocilizumab: a case report, Eur J. Rheumatol 2 (2015) 35–36, https://doi.org/10.5152/eurjrheumatol.2014.14053.
[31] T. Hosoya, F. Mizoguchi, H. Hasegawa, K. Miura, R. Koike, T. Kubota, et al., A case presenting with the clinical characteristics of tumor necrosis factor (TNF) receptor-associated periodic syndrome (TRAPS) without TNFRSF1A mutations successfully treated with tocilizumab, Intern. Med. 54 (2015) 2069–2072, https://doi.org/10.2169/internalmedicine.54.3371.