JETIR.ORG ISSN: 2349-5162 | ESTD Year : 2014 | Monthly Issue
JOURNAL OF EMERGING TECHNOLOGIES AND INNOVATIVE RESEARCH (JETIR)
An International Scholarly Open Access, Peer-reviewed, Refereed Journal

"PFAPA Syndrome in a School-Aged Child: A Case Report Highlighting Diagnostic Challenges in Primary Care"

Ms. Sandeep kaur[1]

Ms. Khushi choudhary[2]

[1]Assistant Professor Maharishi Markandeshwar college of Nursing Maharishi Markandeshwar (Deemed to be) University, Mullana, Ambala PIN: 133203
Email ID-sandynarwal91@gmail.com,
Contact No: 9034379865

[2] Nursing Tutor, Maharishi Markandeshwar college of Nursing Maharishi Markandeshwar (Deemed to be) University, Mullana, Ambala PIN: 133203
Email ID-khushimm09@gmail.com

ABSTRACT

Introduction

PFAPA syndrome (Periodic Fever, Aphthous stomatitis, Pharyngitis, and Adenitis) is the most common cause of periodic fever in childhood. Typically, it occurs in children up to five years old and is characterized by sudden and recurrent episodes of high-spiking fever accompanied by at least one of the other eponymous features. This case report enhances the importance of its recognition, to prevent unnecessary tests and/or treatments.

Case description

We present a case of a Ten-year-old boy, with no relevant background, observed multiple times in primary and secondary care settings due to fever. His parents reported monthly episodes of high fever, usually along with pharyngitis, oral aphthosis, and/or cervical adenitis. He was completely asymptomatic between crises and showed normal growth and psychomotor development. Several oropharyngeal and nasal swab tests were performed, with negative results. After some investigation and articulation with his pediatrician, a diagnosis of PFAPA syndrome was made and he started oral corticosteroids, a single dose on the first day of every episode, obtaining fast and complete symptomatic relief.

Comment

PFAPA syndrome diagnosis is challenging, and its process can trigger anxiety in the patient, his family, and even in healthcare professionals. However, this relatively common and benign condition tends to be self-limited, usually with spontaneous resolution before adolescence. Its recognition by the medical community is essential, particularly in primary care settings since family physicians are usually the first point of medical contact within the healthcare system. This case report enhances the family physician's core competence to manage illness with

nonspecific presentations and the importance of the interface with other specialties to provide the best care to the community.

Keywords: PFAPA syndrome; Periodic fever; Tonsilitis; Cervical adenitis; Aphthous stomatitis; Case report

Introduction

PFAPA syndrome (Periodic Fever, Aphthous Stomatitis, Pharyngitis, and Adenitis) is the most common cause of recurrent fever in children. First described by Marshall et al. in 1987, it is an autoinflammatory condition marked by regular episodes of high fever (39–40 °C), pharyngitis, cervical adenitis, and oral aphthae. These episodes typically last 3–7 days and recur every 2–8 weeks.

Though its exact cause remains unclear, PFAPA is often underdiagnosed. A Norwegian study estimated its incidence at 2.3 per 10,000 children under five. Up to 60% of patients experience prodromal symptoms such as nausea, abdominal discomfort, irritability, headache, and myalgia. Pharyngitis occurs in over 90% of cases, while adenitis is seen in 53–94%, typically involving bilateral, tender anterior cervical nodes. About half of patients develop small aphthous ulcers on non-chewing surfaces of the oral mucosa.

Symptoms usually resolve spontaneously within 4–6 days. Diagnosis is clinical, based on criteria from Marshall and Vanoni et al., but limited awareness often leads to unnecessary tests and ineffective treatments. This case report emphasizes the importance of recognizing PFAPA to support quaternary prevention and avoid medical overuse.

Case description

A 10-year-old boy with no significant family history, allergies, or regular medications was evaluated for recurrent febrile episodes. Since 2019, at age two, he had experienced multiple episodes of high spiking fever (38.5–39.7 °C), frequently accompanied by aphthous stomatitis and cervical adenitis. The initial episode resolved within five days following treatment with paracetamol and nystatin.

Over the following years, the child continued to present monthly with similar symptoms—fever, pharyngitis, aphthous ulcers, and adenitis—prompting repeated visits to primary care and pediatric emergency services. These episodes were often misdiagnosed as tonsillitis or upper respiratory infections, leading to multiple courses of antibiotics with limited benefit.

In May 2025, he presented again with a three-day history of fever and odynophagia. Temperatures exceeded 38.5 °C (peaking at 39.3 °C) and responded only partially to antipyretics. There were no respiratory or gastrointestinal symptoms, and no relevant epidemiological exposures. Examination revealed tonsillar hyperemia and hypertrophy, along with two tender, elastic lymph nodes (~2 cm) in the left anterior cervical chain. No rash or respiratory distress was noted.

Diagnostic workup included negative SARS-CoV-2 and group A Streptococcus swabs, normal urinalysis, and mildly elevated inflammatory markers (ESR: 55 mm/h; CRP: 10.05 mg/dL) with a normal white blood cell count. Serologies for Epstein-Barr virus and cytomegalovirus were negative.

Given the pattern of recurrent, self-limited febrile episodes with associated symptoms, inter-episode wellness, and normal development, a presumptive diagnosis of PFAPA syndrome was made. A single dose of oral betamethasone (0.2 mg/kg) was administered, resulting in rapid symptom resolution, further supporting the diagnosis.

The patient remains under joint follow-up with his family physician and pediatrician. Flares have since become less frequent and are effectively managed with corticosteroids

Comment

PFAPA syndrome, while relatively common in pediatric practice, remains underrecognized among healthcare professionals. Diagnosis is primarily clinical, often guided by the Modified Marshall criteria proposed in 1999.

These criteria, though widely used, are not universally accepted. In 2018, Vanoni et al. introduced a more detailed set of diagnostic criteria that emphasize the nature of fever episodes and the importance of excluding other conditions (see Table 1).

Both criteria sets identify fever as the central symptom, which must be accompanied by at least one of the other hallmark features—aphthous stomatitis, pharyngitis, or cervical adenitis. The Vanoni criteria offer greater specificity, particularly regarding symptom-free intervals and exclusion of other autoinflammatory or infectious diseases.

There are no definitive laboratory tests for PFAPA. However, during flares, mild lymphopenia, neutrophilia, monocytosis, and elevated inflammatory markers such as C-reactive protein may be observed. PFAPA remains a diagnosis of exclusion, requiring careful differentiation from cyclic neutropenia, chronic infections, and immunodeficiencies. In atypical cases, genetic testing for monogenic autoinflammatory syndromes may be warranted

Table 1 Modified Marshall's and Vanoni's criteria set for PFAPA syndrome diagnosis/classification

Criteri set	Modified marshall criteria (1999)	Vanonis criteria (2018)
Age of diseases onset	Before age 5	Typically before age 6
Recurrent fever definition	≥6 episodes of fever lasting 3–6 days, recurring every 2–8 weeks	≥3 episodes of fever lasting 3–7 days, recurring regularly
Symptoms	At least one of: aphthous stomatitis, pharyngitis, cervical adenitis	Fever plus at least one of: aphthous stomatitis, pharyngitis, adenitis
Between flarel periods	Normal growth and development; asymptomatic interval	Symptom-free intervals; normal physical and psychomotor development
Exclusion	No evidence of infection, immunodeficiency, or cyclic neutropenia	Exclusion of infectious, autoimmune, and monogenic autoinflammatory disease

Management

PFAPA syndrome currently lacks standardized, evidence-based treatment guidelines due to the limited number of clinical trials. Management decisions are individualized, based on the severity and frequency of flares, as medical intervention does not appear to alter long-term outcomes.

According to the Childhood Arthritis and Rheumatology Research Alliance PFAPA work group, four main treatment strategies are recognized:

1. Symptomatic relief with antipyretics during episodes
2. Abortive therapy with corticosteroids at flare onset
3. Prophylactic treatment using colchicine or cimetidine
4. Surgical intervention via tonsillectomy

Antipyretics may suffice for milder cases or when parents prefer to avoid corticosteroids or surgery. Corticosteroids are considered first-line abortive therapy, typically administered at the onset of symptoms. Recommended dosing includes:

- Prednisone: 1 mg/kg (max 60 mg), or 2 mg/kg for frequent episodes (≤14-day intervals) or partial response

- Betamethasone: 0.1–0.2 mg/kg orally, as used in this case, with the option to repeat on day two if fever persists

Prophylactic agents like colchicine or cimetidine may be considered in refractory cases, though evidence remains limited. Tonsillectomy is reserved for patients with frequent, severe flares unresponsive to medical therapy

Conflict of interest disclosure

All authors declare that they have no conflicts of interest.

References

1. Amarilyo G, Rothman D, Manthiram K, Edwards KM, Li SC, Marshall GS, et al. Consensus treatment plans for periodic fever, aphthous stomatitis, pharyngitis and adenitis syndrome (PFAPA): a framework to evaluate treatment responses from the childhood arthritis and rheumatology research alliance (CARRA) PFAPA work group. Pediatr Rheumatol Online J. 2020;18(1):31. [Links]

2. Gaggiano C, Rigante D, Sota J, Grosso S, Cantarini L. Treatment options for periodic fever, aphthous stomatitis, pharyngitis, and cervical adenitis (PFAPA) syndrome in children and adults: a narrative review. Clin Rheumatol. 2019;38(1):11-7. [Links]

3. Wang A, Manthiram K, Dedeoglu F, Licameli GR. Periodic fever, aphthous stomatitis, pharyngitis, and adenitis (PFAPA) syndrome: a review. World J Otorhinolaryngol Head Neck Surg. 2021;7(3):166-73. [Links]

4. Vanoni F, Theodoropoulou K, Hofer M. PFAPA syndrome: a review on treatment and outcome. Pediatr Rheumatol Online J. 2016;14(1):38. [Links]

5. Batu ED. Periodic fever, aphthous stomatitis, pharyngitis, and cervical adenitis (PFAPA) syndrome: main features and an algorithm for clinical practice. Rheumatol Int. 2019;39(6):957-70. [Links]

6. Førsvoll J, Kristoffersen EK, Øymar K. Incidence, clinical characteristics and outcome in Norwegian children with periodic fever, aphthous stomatitis, pharyngitis and cervical adenitis syndrome: a population-based study. Acta Paediatr. 2013;102(2):187-92. [Links]

7. Thomas KT, Feder HM Jr, Lawton AR, Edwards KM. Periodic fever syndrome in children. J Pediatr. 1999;135(1):15-21. [Links]

8. Kutsuna S, Ohmagari N, Tanizaki R, Hagino N, Nishikomori R, Ujiie M, et al. The first case of adult-onset PFAPA syndrome in Japan. Mod Rheumatol. 2016;26(2):286-7. [Links]

9. Gattorno M, Sormani MP, D'Osualdo A, Pelagatti MA, Caroli F, Federici S, et al. A diagnostic score for molecular analysis of hereditary autoinflammatory syndromes with periodic fever in children. Arthritis Rheum. 2008;58(6):1823-32. [Links]

10. Soon GS, Laxer RM. Approach to recurrent fever in childhood. Can Fam Physician. 2017;63(10):756-62. [Links]

11. Padeh S, Stoffman N, Berkun Y. Periodic fever accompanied by aphthous stomatitis, pharyngitis and cervical adenitis syndrome (PFAPA syndrome) in adults. Isr Med Assoc J. 2008;10(5):358-60. [Links]

12. Wurster VM, Carlucci JG, Feder HM Jr, Edwards KM. Long-term follow-up of children with periodic fever, aphthous stomatitis, pharyngitis, and cervical adenitis syndrome. J Pediatr. 2011;159(6):958-64. [Links]