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DEMANDING DIFFERENTIAL DIAGNOSIS OF "UNCLEAR ABDOMINAL ILLNESS" WITH POSSIBLE SURGICAL CONSEQUENCE - FAMILIAL MEDITERRANEAN FEVER (FMF)

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Introduction

- The differential diagnosis of 'unclear / acute abdomen' is widespread.
- In addition to the most common clinical symptoms & diseases, 'exotic Dx' are also occasionally of importance.
- As a result of increasing global migration, rare diseases of certain origins are important differential diagnoses for unclear & recurrent symptoms.
- The importance of the medical history as an immensely important pillar of clinical practice becomes clear.
- Even with typical clinical signs of appendicitis may conceal other diseases.

Objective

The following "case report" should put the light on the interesting & rarely described case of a patient with 'Fam. Mediterranean fever' as a rare differential diagnosis of "unclear abdomen" in Germany.

CASE DESCRIPTION (1) - medical history

➢ **Current (present) medical history:**
- Male patient (19 yrs)
- Presentation with fever since 2 d
- Abdominal pain, joint pain & breathing-dependent pulling chest pain
- Such symptoms - more frequent recently but had disappeared on their own after 2 d
- Travelling to tropical regions in the last 6 months denied

➢ **Medical history:**
- No home medication
- No previous illnesses
- Noxas: * Non-smoker
  * No alcohol consumption

CASE DESCRIPTION (2) - Diagnostics

➢ **Vital signs:**
AF: 18/min, HR: 100/min, Temp.: 37.5°C

➢ **Physical examination:**
- Patient in reduced general condition & normosomic nutritional status
- Lying in bed with bent legs,
- Neurological orientation unremarkable,
- Cardiopulmonarily stable

➢ **Examination of the abdomen:**
- Scanty bowel sounds over all 4 quadrants
- Defensive tension with generalised pressure pain (interpreted as peritoneal irritation),
- Pain: strongest in the right lower quadrant, McBurney-test (+) // Lanz-test (+) // Psoas sign (-)
- Complaint of concussion pain
- Adopts protective posture

➢ **Laboratory parameters:**
* White blood cell count: 8.9; CRP: 35 (SI)
* Remaining laboratory (incl. liver & kidney values)
  → unremarkable

➢ **Ultrasound:** → no pathological findings

CASE DESCRIPTION (3) - clinical course

➢ **Under intravenous pain medication with Metamizole:**
- Clear improvement - decrease of fever
- Defense tension persists during abdominal examination

➢ **Continuation of pain medication & laboratory checks**

➢ **Extended family history:**
- Parents are related
- A relative in the family with similar symptoms
  → recurrent fever attacks with generalised abdominal pain for 2 years

CASE DESCRIPTION (4) - clinical course after 3 days

➢ **Patient free of fever & symptoms, feels perfectly healthy again & would like to go home**

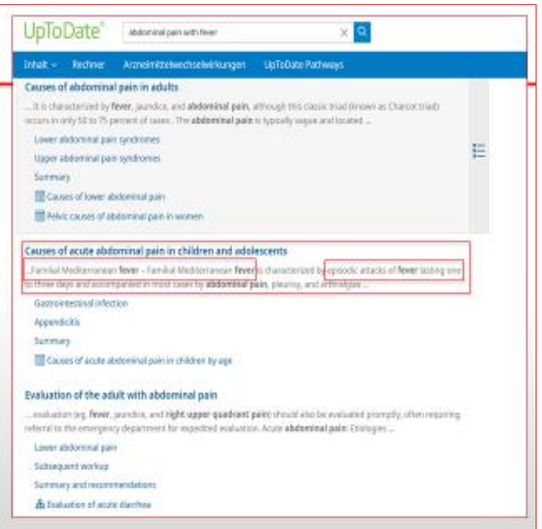
➢ **Laboratory:**
Inflammation values within the normal range

➢ **What else could be done to obtain the diagnosis?**

➢ **Medical history:**
→ **Indications of a familial or genetic component!**

FMF

- Search in UpToDate for: 'abdominal pain & fever '
- If considering the familial component, possible indication of



Familial Mediterranean Fever (FMF)

General information

First described in 1908 (JANEWAY *et* MOSENTHAL), name by HELLER 1955 other names, e.g., familial recurrent polyserositis

Genetics & etiology

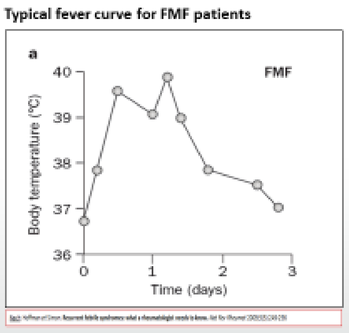
Familial Mediterranean Fever Gene (MEFV) already described in 1997 Autosomal recessive inheritance

Chromosome 16 (gene locus 16p13.3), mutations in all exons, often exon 10 (694 & 680) & then exon 2 (148)

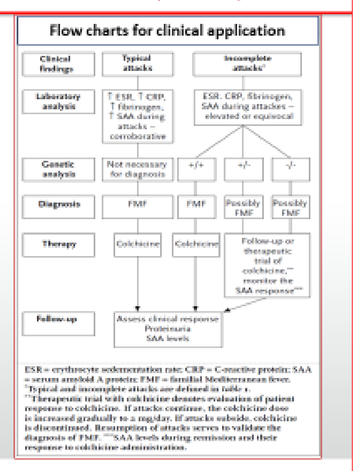
→ Hyperactivity of neutrophil granulocytes resulting in spontaneous autoinflammation (inflammasome & IL-1)

Familial Mediterranean Fever (FMF)

Typical fever curve for FMF patients



Lidar M, Livneh A.
Familial Mediterranean fever: clinical, molecular & management advancements.
Neth J Med 2007;65(9):318–324



Diagnostic criteria according to Tel HASHOMER

Familial Mediterranean Fever (FMF) ... or more simply for clinical practice

Figure 1. Simplified Tel-Hashomer Criteria for the diagnosis of FMF.[2]

Major criteria	Minor criteria
1. Recurrent febrile attacks accompanied by peritonitis, synovitis or pleuritis. 2. Amyloidosis of the AA-type without predisposing disease. 3. Favourable response to continuous colchicine treatment.	1. Recurrent febrile attacks. 2. Erysipelas-like erythema 3. FMF in a first degree relative

Acc. to: Cliff-Patel *et al.* **Familial Mediterranean fever: a differential diagnosis for the surgical abdomen.** JRSM Open2022;13(9):20542704221123433

Conclusion

Familial Mediterranean Fever (FMF)

➢ Family & social aspects of medical history are important
➢ Consider rarer familial syndromes
➢ Consider the diagnosis of FMF with origin from the Mediterranean region after exclusion of acute diseases
➢ Apply Tel-HASHERMO criteria & determine amyloid A in serum (SAA) in acute flare-ups
➢ Therapy of choice → Colchicine
➢ Information about complications if therapy is not implemented → Amyloidosis (kidney, heart) can be considered strongest survival-determining factor!

References

- UpToDate: **Clinical manifestations and diagnosis of familial Mediterranean fever** (access, 10/03/2022)
- UpToDate: **Causes of abdominal pain in adults** (access, 10/03/2022)
- UpToDate: **Causes of acute abdominal pain in children and adolescents** (access, 10/06/2022)
- UoToDate: **Tumor necrosis factor receptor-1 associated periodic syndrome (TRAPS)** (access, 12/04/2022)
- Orphanet: Familial Mediterranean fever (access, 10/08/2022)
- Cliff-Patel *et al.* **Familial Mediterranean fever: a differential diagnosis for the surgical abdomen.** *JRSM Open* 2022;13(9):20542704221123433
- Tufan *et al.* **Familial Mediterranean fever, from pathogenesis to treatment: a contemporary review.** *Turk J Med Sci* 2020;50(SI-2):1591-1610
- Hoffman HM, Simon A. **Recurrent febrile syndromes: what a rheumatologist needs to know.** *Nat Rev Rheumatol* 2009;5(5):249-256
- Lidar M, Livneh A. **Familial Mediterranean fever: clinical, molecular and management advancements.** *Neth J Med* 2007;65(9):318–324