Misnomer Leading to Delayed Diagnosis of an Autoinflammatory Syndrome in a Latino Male

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Introduction

- Familial Mediterranean Fever (FMF)
  - Relatively rare auto-inflammatory disease
  - Autosomal recessive inheritance pattern
  - Ordinarily found in patients of Mediterranean and Middle Eastern descent, however it can affect any ethnic group
  - Symptoms are non-specific and mimic many common diseases, leading to
    - Unnecessary workup
    - Exploratory surgery
    - Delayed diagnosis

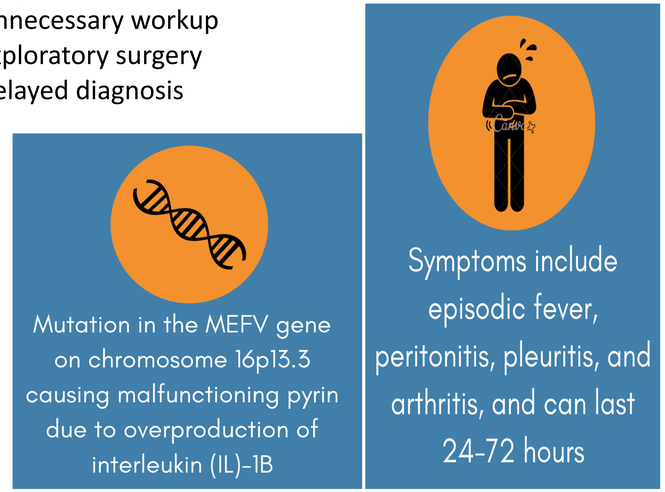

Figure 1: Genetics and symptoms of FMF

Figure 2: Most common populations affected by FMF

- We describe a patient with a classic presentation of FMF, which was overlooked due to implicit bias, racial disparities, and a disease misnomer

Case Presentation

- 30-year-old Latino male with past medical history of appendectomy and "IBS" who presents with:
  - Cyclical episodes of peritoneal abdominal pain for 15 years
    - Pain described as severe and is unable to get out of bed or walk without hunching over
  - Associated with constipation, bloating, chills, night sweats
  - No associated chest pain, joint pain, or rashes
- Past Medical History and Historical Timeline:
  - First attack at age 9
  - Attacks occurred twice per year, lasting 3-4 days, and severity was significantly exacerbated by stress
  - Tried laxatives and prune juice for constipation
  - Family history is significant for multiple members in Mexico diagnosed with with irritable bowel syndrome (IBS) or *"colitis nerviosa"*
    - His parents are originally from Jalisco, Mexico, an area that was heavily colonized by the Spanish
  - Patient receives a diagnosis of IBS-C, even though his constipation would improve between attacks
  - Age 10: underwent colonoscopy which was normal and then decision was made for appendectomy
  - He continued to undergo multiple CT scans and even a small bowel series which were unrevealing
  - All labs were normal

Workup and Clinical Course

- His lab work and physical exam were unremarkable between attacks
- Patient did not meet Rome criteria for IBS and due to suspicion for recurrent peritonitis, FMF mutation testing was ordered, and this revealed that the patient was heterozygous for M694I mutation
- Due to the classic clinical presentation and supportive genetic testing, he was started on 1.2 mg of Colchicine daily
- At 6 months follow up, he has not had any further attacks

Discussion

- FMF can affect any ethnic group, making the disease name a misnomer
  - Carrier rate and the severity of disease manifestation vary considerably both among and within different ethnic groups
  - New populations FMF has been identified in include: Anglo-Saxons and Germans, British, Afghans, Indians and Chinese, Italians, Spanish, French, Greeks and Cypriots, Japanese
- One study describes 52 patients diagnosed with FMF living in Mexico City, 39 of which were born there
  - The predominant features were fever and typical peritoneal type abdominal pain
  - 47 surgeries were performed (exploratory laparotomy), 24 underwent one surgery, 20 two surgeries, and 3 three surgeries each
- In Mexico, there are families with Mediterranean ancestors that have spread the disease to second and third generation offspring already born in Mexico

Conclusions

- This case illustrates the implications of a misnomer as well as how racial disparities and implicit bias delayed diagnosis in a patient with a classic presentation of Familial Mediterranean Fever
- The patient was untreated for over two decades and may have undergone an unnecessary appendectomy
- It is important to remember that despite its name, Familial Mediterranean Fever can affect any ethnic group

References

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