Not Quite Bronze: A Fiery Rash Reveals Hidden Hemochromatosis

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- Abstract:
- Hereditary Hemochromatosis is an autosomal recessive genetic disorder characterized by excessive iron accumulation in tissues. Classic early symptoms, including fatigue, arthralgia, and hyperpigmentation, typically begin in the 4th decade of life. This case highlights a 29-year-old male with an unusual presentation of Hereditary Hemochromatosis.
- A 29-year-old Caucasian male presented to his primary care provider (PCP) with complaints of an erythematous and pruritic rash that was present across his chest and abdomen, and dry, scaly hands. He was treated with steroids with temporary improvement. 2 weeks later, the patient visited the emergency department, presenting with a beefy, red, and pustular rash diffusely across both feet. Due to recent irritant exposure to new laundry detergent and sawdust, his rash was assumed to be eczematous, and he was given steroids and antibiotics due to a few open sores observed. Upon returning to his PCP for follow up, routine labs revealed an elevated iron level, at 208. In conjunction with his skin irritation, a cheek swab was ordered to check for the C282Y gene. This prompted his new diagnosis of Hereditary Hemochromatosis (HH). The patient was referred to hematology for further investigation. MRI of the abdomen revealed diffuse iron deposition in the liver. Weekly phlebotomy was started to reduce total body iron stores. This case is uniquely seen in a 29-year-old, whereas the median age of males diagnosed with hemochromatosis is 40-60 years old. The atypical presentation of recurrent dermatitis, most recently with the rash on both feet, is also a distinctive finding.
- Recognizing atypical presentations of Hereditary Hemochromatosis may facilitate earlier diagnosis and treatment, potentially preventing long-term complications such as liver cirrhosis, diabetes mellitus and dilated cardiomyopathy. Physicians should consider iron studies in patients with eczema or irritant dermatitis that are persistent despite standard therapies.

Introduction

- Hereditary hemochromatosis (HH) is a genetic disorder in which the body absorbs and stores excess iron, resulting in iron overload and deposition of iron throughout the body [1].
- Hemochromatosis presents more frequently in males, typically between the 4th and 6th decades[1].
- The most common cause is a homozygous C282Y mutation in the HFE gene, resulting in dysregulation of the protein hepcidin. This causes an increase in iron uptake from the diet and release from macrophages and hepatocytes [2].
- The classic triad of hemochromatosis is skin hyperpigmentation, cirrhosis, and diabetes mellitus[3]. This presentation contributes to the popular moniker given to hemochromatosis of “bronze diabetes.”
- Although most patients are asymptomatic initially, iron deposition across organ systems can lead to a wide array of presentations such as hypothyroidism, xerosis, arrhythmias, cardiomyopathy, arthralgias, abdominal pain, hepatomegaly, jaundice, and amenorrhea [5].
- Patients are at risk for liver cirrhosis, hepatocellular carcinoma, and hepatic encephalopathy [6].
- Dermatologic manifestations have yet to be thoroughly studied in their various presentations [7]

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Methodology

- A 29-year-old Caucasian male with no significant medical history presented to his primary care provider with complaints of fatigue and an erythematous macular pruritic rash.
- He reported that this rash had been present across his chest and abdomen persistently for three months. It was initially attributed to irritant contact dermatitis, given that his rash was on his abdomen, near a nickel belt buckle. His frequent history of urgent care visits in 2018 also corroborated this diagnosis. Fatigue preceded this rash, and he also endorsed mild arthralgias.
- Two weeks later, the patient visited the emergency department, presenting with a more extensive rash. The rash featured an erythematous base with numerous pustules diffusely across the dorsum of both feet, as shown in Figure 1.
- Upon follow-up with his primary care physician, routine labs were ordered to explore alternative causes, revealing an abnormal iron panel shown in Table 1. His skin irritation, combined with fatigue and an abnormal iron panel led the provider to order a cheek swab to check for the C282Y gene. The finding of a homozygous mutation in the HFE gene confirmed his new diagnosis of HH.
- Upon this result, he was referred to hematology for further work-up. The iron panel was once again abnormal with the patient’s values shown in Table 1. Figure 2 shows the magnetic resonance imaging (MRI) of the abdomen demonstrating diffuse signal loss on T2* imaging consistent with diffuse iron deposition in the liver.

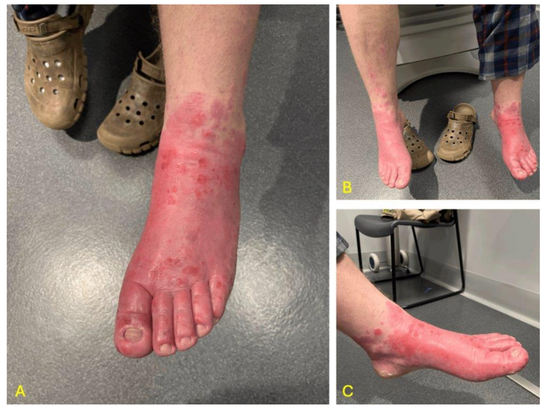

Figure 1: *Images of novel rash across both feet*
(A): Anterior view of the right foot showing a diffuse erythematous rash with numerous pustules. (B): Wide anterior view of both feet demonstrating a diffuse erythematous rash with multiple pustules. (C): Medial view of the right foot highlighting the diffuse erythematous rash.

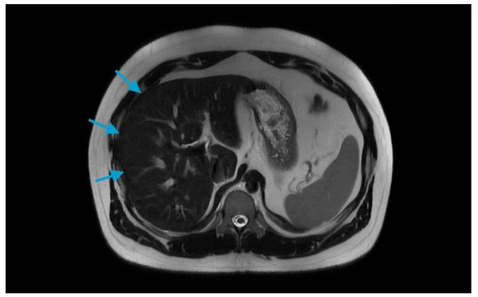

Figure 2: Diffuse signal loss on T2 imaging (arrows) throughout the liver is present with evidence of signal increase on opposed gradient echo imaging indicating diffuse sign infiltration.

Iron panel	Patient values 11/12/2024	Patient values 02/10/2025	Reference range
Serum iron	208 mcg/dL	108 mcg/dL	60–170 mcg/dL
Ferritin	719 ng/mL	473.4 ng/mL	24–336 ng/mL
Transferrin saturation	75%	40%	20–50%
Total iron binding capacity	279 mcg/dL	269 mcg/dL	250–450 mcg/dL
Transferrin	–	192 mg/dL	215–380 mg/dL

Table 1: Iron studies from initial PCP visit and recent hematology visit.

Results

- A unique characteristic about this case is the patient's age. Our patient was diagnosed with type 1 HH at the age of 29, and he had recurrent cutaneous symptoms for several years prior to diagnosis.
- Our patient had characteristics of classic adult-onset hemochromatosis, with his slow progressive onset of vague fatigue, arthralgia, and rashes.
- Juvenile HH on the other hand is typically a rapid onset disease process presenting earlier in life with severe features such as cardiomyopathy, hypogonadism, and cirrhosis [8].
- Hemochromatosis can be difficult to diagnose as it often presents as nonspecific symptoms in its early stages. Therefore, genetic testing remains the gold standard for diagnosis [9].
- Given that the mainstay treatment of hemochromatosis, phlebotomy, is inexpensive and easy to access, there should be a low threshold to test patients who have signs of iron overload.
- In this case report, we discussed a patient who presented with recurrent episodes of resistant dermatitis for years. This nonspecific rash masquerading as recurrent dermatitis fostered a delay in diagnosis and treatment due to the indistinct presentation.
- Phlebotomy typically improves skin hyperpigmentation but given this patient's atypical rash, it is unclear whether reducing iron load would fully reverse the damage.

Conclusion

- This case highlights an initial presentation that deviates from the typical findings and age group seen in hemochromatosis.
- Recognizing such manifestations may facilitate earlier diagnosis and treatment, potentially preventing long-term complications such as liver cirrhosis, diabetes mellitus, and dilated cardiomyopathy.
- Since treatment is accessible and effective, providers should consider iron studies in patients with resistant dermatitis that persists despite standard therapies.