A pigmented octogenarian

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Abstract

An 81-year-old woman presented with cachexia, malaise, deranged liver function tests (LFTs), hepatomegaly and hyperpigmentation. Hereditary haemochromatosis (HH) was ultimately diagnosed on genetic testing. Haemochromatosis is a difficult diagnosis in older people because not only are the symptoms protean and occur commonly in other geriatric conditions, but HH is also considered an unlikely new diagnosis in the over 80s; it is generally felt to present in the 5th decade for men or 6th decade for women. However, older people can present with only mild disease, so diagnosis is important not just for family screening but also since treatment can reduce unnecessary morbidity and mortality in a population with increasing longevity.

Keywords: Hereditary haemochromatosis, hyperpigmentation, ferritin, elderly, older people

Case report

An independent 81-year-old woman was referred by her general practitioner with cachexia, malaise, increasing leg oedema, worsening knee pain and deranged LFTs. Her past medical history included a partial colectomy for sigmoid cancer, treated hypothyroidism, osteoarthritis and a permanent pacemaker. Her regular medications were aspirin, levothyroxine, folate, ranitidine and co-amilofruse. She lived alone, walked with a stick, was a lifelong non-smoker and drank occasional alcohol.

Positive examination findings included bibasal crepitations, bilateral pitting oedema to the knees and smooth hepatomegaly. She was only later noted to appear tanned, despite spending little time outdoors. LFTs at presentation were ALT 108, ALP 343 and bilirubin 17. All other routine bloods were normal.

Initial diagnosis was of heart failure. With treatment, the patient’s symptoms improved. The pacemaker check was normal, and echocardiogram showed reasonable left ventricular function.

However, there was concern about underlying malignancy, so a computer tomography of the chest/abdomen/pelvis was done. Lesions were found in the right lung apex and the uterus. The chest lesion was minor and inaccessible to biopsy, but she underwent an examination under anaesthetic by the gynaecologists, which was unremarkable. She had negative tumour markers, and normal synacthen test and liver screen — apart from a ferritin of 3231.

Iron studies revealed a transferrin saturation of 74%. Genetic testing confirmed a diagnosis of HH, showing she was homozygous for the C282Y mutation.

The patient was discharged home and after declining liver biopsy, underwent a successful programme of slow venesection reducing her transferrin saturation to below 50%. Screening was arranged for her family.

Discussion

HH is a common autosomal recessive derangement of iron metabolism, with clinical manifestations related to iron deposition in tissues. Peak incidence is between 40 and 60 years with women presenting later due to therapeutic menstrual loss. Early symptoms are non-specific: fatigue, weakness, arthralgia and abdominal pain. Even later complications such as liver disease, diabetes, cardiac problems and impotence do not always present together. Thus the diagnosis is not straightforward in the elderly with multiple comorbidities.

Symptomatic haemochromatosis is usually associated with homozygosity for the C282Y mutation of the HFE gene. However, not all homozygotes develop iron overload.
Estimates suggest that 70,000 people in the UK have symptomatic HH; only about 3,000 cases are recognised [1]. This may be due to onset being later than previously believed [2] and variable phenotypic penetrance in homozygotes; one study has suggested a penetrance of <1% [3]. This raises issues for population screening.

Optimal treatment involves regular venesection, which is not without pitfalls in older people, who often have other co-morbidities and struggle with the logistics of frequent hospital visits [4].

Treatment prevents progression to cirrhosis in pre-cirrhotics and also the onset of non-hepatic complications. Some symptoms improve but there is no effect on diabetes, arthritis or impotence. The presence of cirrhosis dramatically reduces prognosis, and treatment can only slow progression to liver failure; it has no impact on the incidence of hepatocellular carcinoma which causes death in 30% of patients [5].

Since older patients may still only have mild disease at presentation, HH is an important diagnosis to consider. This is not only for screening family members, but also because early intervention can decrease morbidity in a group whose life expectancy has increased over the last 50 years.

Key points

- HH is not commonly diagnosed in the elderly; it may often be missed due to its multifaceted presentation mimicking common geriatric conditions.
- Older people may present with only mild disease and therefore gain significant benefit from treatment.

Conflict of interest

None.

Informed consent

The patient discussed in this report has given written consent for using details of her case for purposes of medical education, including publication in a scientific journal.

References


Severe exfoliative dermatitis caused by strontium ranelate: two cases of a new drug reaction

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Abstract

Strontium ranelate is a relatively new drug used as a second-line treatment for osteoporosis, often targeted at older patients. It is known to cause skin rash and rarely drug reaction with eosinophilia and systemic symptoms, but there are no reports of exfoliative dermatitis as a reaction in the literature. We present the first two cases of this adverse effect of the drug, combined with