Coexistence of iron metabolism disorders in patients with known Inherited Metabolic Diseases.

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BACKGROUND
Iron metabolism involves a series of complex biochemical processes. Although it is rare, primary iron overload disorders may co-exist with other Inherited Metabolic Diseases (IMDs) and impact upon their management.

It is worth considering coexisting diagnoses in patients already affected with IMDs. In some cases, deranged iron studies may be the first indication to General Practitioners and hospital clinicians of a possible underlying IMD.

METHODS
This involved a retrospective review of patients with IMD in whom disorders of iron metabolism were also identified.

RESULTS
Three adult patients affected with Classical Galactosaemia (M32), Phenylketonuria (PKU; M34) and MELAS (F45) were found to have abnormal biochemical parameters of iron metabolism: ferritin, iron saturation, total iron binding capacity, full blood count and serum iron. All three patients were later diagnosed with Haemochromatosis in adult life and required regular venesection. A low-protein diet with synthetic amino acid supplements was needed in the patient with PKU, which contains variable amounts of iron (5.3-11.8mg); this may affect iron studies indices.

In a further patient without a known diagnosis of IMD, deranged iron studies and liver function tests (LFTs) led to a diagnosis of aceruloplasminaemia in adolescence.

DISCUSSION
It is important to assess iron studies as part of a nutritional screen in patients with IMD, in particular in the context of abnormal LFTs.

It is worth noting IMD dietary supplements may affect biochemical parameters of iron metabolism. A multidisciplinary approach with involvement from hepatology, haematology and the metabolic team is recommended in such cases involving a dual diagnosis.

<table>
<thead>
<tr>
<th>Parameter</th>
<th>Expected derangement</th>
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<tr>
<td>Serum iron</td>
<td>Raised (may be normal owing to biological variability)</td>
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<tr>
<td>Transferrin saturation</td>
<td>Raised (&gt;50% males, &gt;40% females)</td>
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<tr>
<td>Total iron binding capacity</td>
<td>Low</td>
</tr>
<tr>
<td>Ferritin</td>
<td>Raised (&gt;300 ug/l males, &gt;200 ug/l females)</td>
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Table 1. Expected biochemical findings when measuring iron studies and ferritin in those with hereditary haemochromatosis (based on a northern European population)

CONCLUSION
This small number of cases highlights that in patients with known IMD, it is important to consider the possibility of co-existing disorders such as disorders of iron overload. Including iron studies as part of a nutritional screen in patients with IMD will improve the identification of such disorders, leading to earlier identification and improved outcomes for such patients.

Conflicts of interest: None

REFERENCES