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Pyridoxine-sensitive X-linked 'sideroblastic' anaemia in the absence of ring sideroblasts - the molecular diagnosis of congenital red cell disorders

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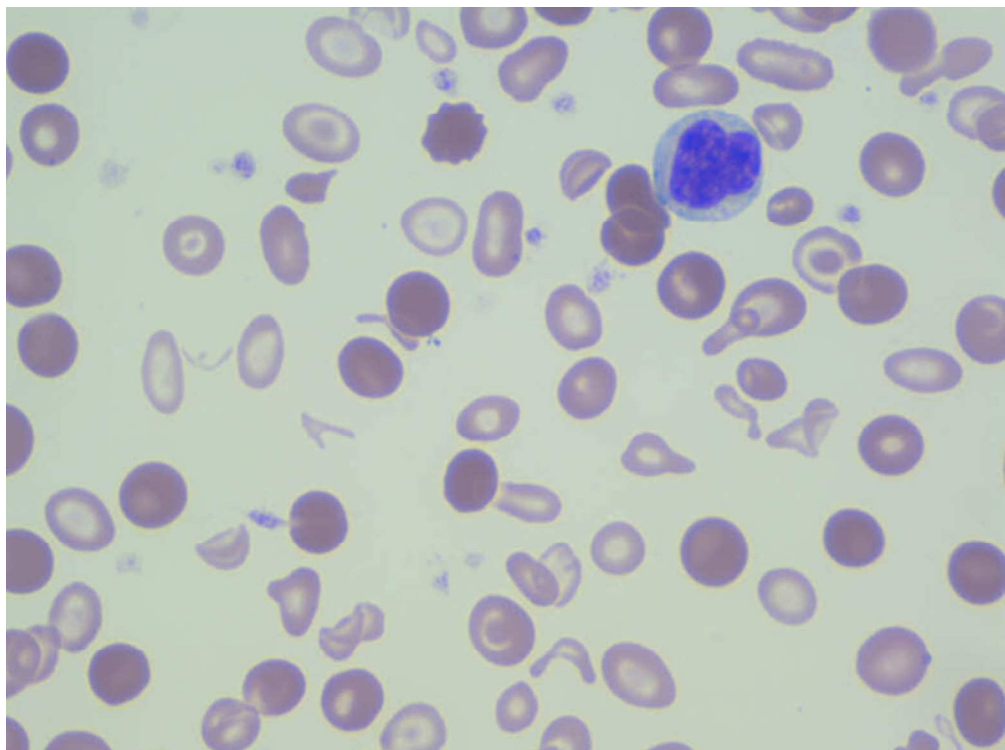


Figure 1: Peripheral blood
57x42mm (300 x 300 DPI)

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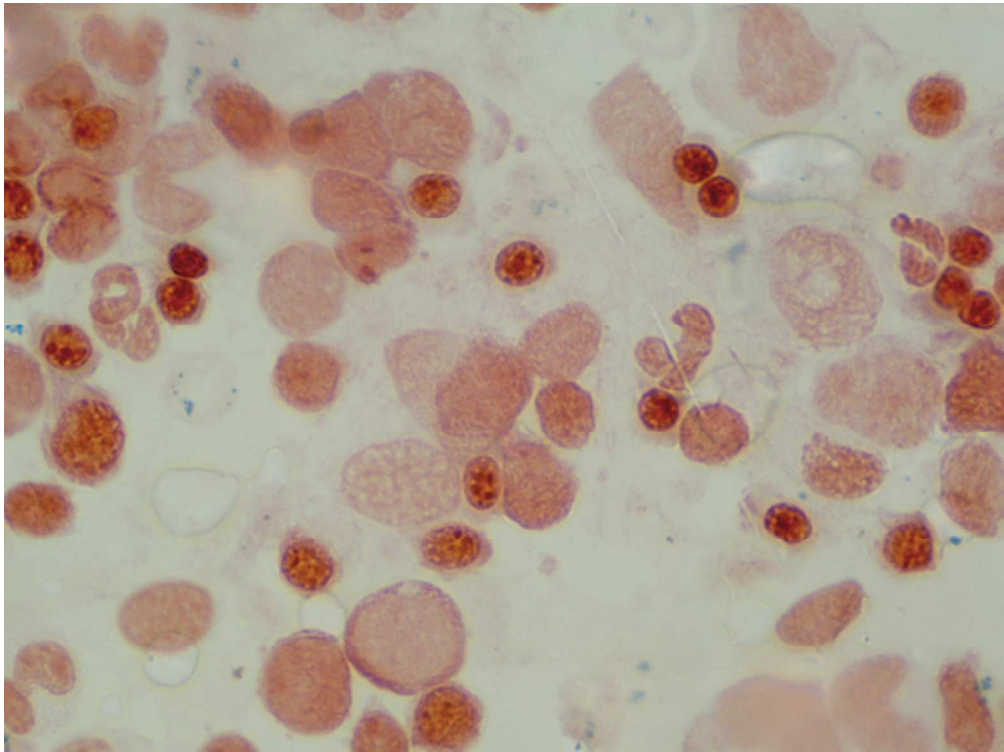


Figure 2: Bone marrow aspirate (Perls' stain)

57x42mm (300 x 300 DPI)

review

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3 **Pyridoxine-sensitive X-linked 'sideroblastic' anaemia in the absence of ring**
4 **sideroblasts – the molecular diagnosis of congenital red cell disorders**
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10 Clark², and Dr Peter Carey¹
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19 Foundation Trust, Newcastle-upon-TyneUK
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16 His full blood count showed a haemoglobin concentration (Hb) of 27 g/l, MCV 48 fl and
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18 MCH 13.5 pg, with reticulocytes $0.40 \times 10^9/l$. White cell count was $6.62 \times 10^9/l$, neutrophil
19
20 count $2.23 \times 10^9/l$, platelet count $311 \times 10^9/l$ and ferritin 262 $\mu g/l$. A peripheral blood film
21
22 demonstrated dimorphic red cells with one population of normocytic, normochromic
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24 cells and a second population with marked poikilocytosis, target cells, irregular
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Pyridoxine-sensitive X-linked 'sideroblastic' anaemia in the absence of ring sideroblasts – the molecular diagnosis of congenital red cell disorders

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