DELAYED DIAGNOSIS OF HAEMOPHILIA IN A CHILD WITH BRUISING

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ABSTRACT

Bruising in small children can give diagnostic challenges (1-3). We present a case in whom the diagnosis of Haemophilia A was delayed by a year. Increased vigilance might have allowed this diagnosis to have been made earlier.

KEYWORDS

Haemophilia, bruising, non-accidental injury, child safeguarding, awareness

CASE REPORT

A 2 year old boy of Eastern European parents was referred to social services by concerned nursery staff who had observed multiple bruises for which they could find no explanation. They referred him to Community Paediatricians for further assessment. The mother reported that she had observed a similar pattern of bruises when her son was a year of age. She had been concerned at the time and took him to the general practitioner who had reassured her that this level and type of bruising was normal in a child learning to walk. No investigations had been carried out at this initial presentation. The mother reported that her son was an active boy who frequently falls and bruises. He did not bruise as an infant, prior to becoming mobile. There were no antenatal or postnatal concerns, including excessive bleeding at the time of birth. The child had a normal developmental trajectory and his immunizations were up to date. No bleeding or bruising had been observed after the administration of vaccines. There had been no episodes of bleeding into mucosae, muscles or joints. The family history was unremarkable; specifically there was no history of bruising, bleeding or joint swellings.

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On examination the child appeared well, stable with no dysmorphic features. He had multiple symmetrical greenish coloured bruises measuring from a few centimetres to maximum of 2.7 centimetres (see figure 1). These were distributed over the trunk, shins, lower abdomen and lower arms and were approximately equal in number on both sides of the child’s body. There was no bruising around the ears or neck, that is, within the ‘safe triangle’. There was no bruising found on the scalp, palmar or plantar surfaces. There was no bruising on the cheeks of his face. A systemic examination was normal, including neurological examination. There was no bleeding evident from any site. Investigations showed a Haemoglobin of 9.9 g/dl, with a normal platelet count and film. Renal and liver functions were normal. Clotting factor checks gave the diagnosis of Haemophilia A within 24 hours of presentation. The child has been treated in collaboration with a national haemophilia centre; the family has been reviewed by a geneticist. Social services did not identify any risk factors in the care of the child.

Abnormal results from investigations in this case:

<table>
<thead>
<tr>
<th>Test</th>
<th>Normal range</th>
<th>Patient</th>
</tr>
</thead>
<tbody>
<tr>
<td>Prothrombin time</td>
<td>12 - 14 secs</td>
<td>10.3*</td>
</tr>
<tr>
<td>Activated partial thromboplastin time</td>
<td>30 - 45 secs</td>
<td>41.6</td>
</tr>
<tr>
<td>Partial Thromboplastin time</td>
<td>30 - 50 secs</td>
<td>81.1*</td>
</tr>
<tr>
<td>Thrombin time</td>
<td>10 - 13 secs</td>
<td>14.1*</td>
</tr>
<tr>
<td>Factor VIII</td>
<td>50 - 150 IU/dL</td>
<td>&lt;1.0*</td>
</tr>
</tbody>
</table>

**DISCUSSION**

Children presenting with unexplained bruises require blood tests to exclude an underlying bleeding disorder. Bruising in children may indicate inappropriate handling or abuse (1-3). The skin is the most commonly damaged organ that can be visualized in abused children. Further, post-mortem studies suggest bruising may be an indicator or flag for potentially significant damage to a child by its carers or someone in the immediate family home (4). Non-accidental injury is more common than underlying bleeding disorders. However the Royal College of Paediatrics and Child Health, as well as the National Institute of Health and Clinical Excellence recommend investigation as in some cases, as in that presented here, a propensity to bruise may be revealed by the daily traumas of toddler life. Further, bruises cannot
be dated accurately and relatively few patterns help distinguish non-accidental from accidental injury (5,6)

Figure 1
The haemophilias are sex-linked recessive bleeding disorders with a partial or complete lack of certain clotting factors. Haemophilia A is secondary to deficiency of factor 8. Haemophilia B relates to a deficiency of factor 9. In the United Kingdom it is estimated there are 6000 cases of haemophilia, of whom 90% have haemophilia A. The Centre for Disease Control and Prevention (CDC) reported that the median age at diagnosis is 36 months for mild hemophilia, 8 months for moderate hemophilia and 1 month for severe haemophilia. In a ten year retrospective survey of haemophilic children in Kansas, it was found that 15.3% had been initially suspected of being abused because of the bruises they showed on first presentation. In this cohort von Willebrand disease was found as frequently as haemophilia (6). The observations in this case are consistent with these.

Nearly a third of cases of haemophilia have no previous known family history; cases may be identified in all racial groups. Some arise from de novo mutations and variable penetrance is well known. Variations in factor levels – the ecology of the clotting system - may manifest in delayed or intermittent presentations (7-9). More severe cases of bleeding are typically also accompanied by thrombocytopenia (10). Our case had presented to GP in the first year of life, but was not formally investigated so that an opportunity to make an appropriate diagnosis was missed. From the perspective of more severe presentations it has been observed by national haemophilia centres that primary care doctors, paediatricians and emergency departments need to be made aware of the diagnosis of clotting disorders as a cause of unexplained bruising or bleeding (7-9).

CONCLUSION

It is important to exclude anomalies of the clotting process in children presenting with unexplained bruises or other bleeding, as expeditiously as possible. Delays in diagnosis may result in a more severe presentation (10). Both primary and secondary services need to maintain a high index of suspicion both of possible underlying bleeding disorders and non-accidental injury. A thorough history, meticulous examination and investigation of checks of the platelets, clotting factors and the connective tissues are required in order to correctly identify clotting disorders in children.

REFERENCES

[4] Ingham AI, Langlois NE, Byard RW. The significance of bruising in infants - a


