Combined Factor V and VIII Deficiency

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Dear Editor,

This is the first report of an Omani family with combined factor V and VIII deficiency. The family has six members: the parents, three affected children, and one unaffected child. The first affected member was a 24-year-old unmarried Omani female (Case A) who had a history of recurrent minor gum bleeding while tooth brushing and bruises with minor trauma, but no history of heavy menses. She had a medical history of heavy bleeding at the age of one week after female circumcision for which she received three units of blood. At presentation, her complete blood count was normal. The coagulation screen revealed a prothrombin time (PT) of 16.8 s (normal range (NR): 9.9–11.9), activated partial thromboplastin time (APTT) of 80.1 s (NR: 26.5–37.5) and thrombin time (TT) of 17.7 s (NR: 12.8–17.6). The coagulation factors assay revealed low factor V (0.101 u/mL) and VIII (0.126 u/mL; chromogenic assay).

Two cases (B and C) were found to have combined factor V and VIII deficiency. The physical examinations and complete blood counts of cases B and C were normal. The coagulation screen for case B revealed a PT of 16.5 s, an APTT of 78.5 s, and a TT of 18.0 s. Factor V and VIII levels were 0.099 u/mL and 0.193 u/mL, respectively. Similarly, the coagulation screen for case C revealed a PT of 16.5 s, an APTT of 79.9 s, and a TT of 17.7 s with factor V and VIII levels of 0.102 u/mL and 0.067 u/mL, respectively. The 12-year-old sister and both parents were clinically well with normal coagulation screens.

A diagnosis of familial multiple factor deficiency Type 1 was made based on prolongation of both PT and APTT with low levels of factor V and VIII. This is an exceedingly rare disorder, [1–3] and not previously described in Oman. It is due to a mutation in the trafficking of factors V and VIII to outside the cell.[1,4] Although this is very rare, we recommend testing for these factors in Omani patients with prolonged PT and APTT. These rare coagulation defects are usually recessive,[2] and in a culture where consanguinity is high the probability of affected families is higher. This report also documents its existence in Oman.

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