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Case Report

A Pregnancy Complicated with Fechtner Syndrome: A Case Report

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Fukada, Y., Yasumizu, T., Sumino, E. and Hoshi, K. *A Pregnancy Complicated with Fechtner Syndrome: A Case Report.* Tohoku J. Exp. Med., 2000, **191** (3), 183-186 —— A 21-year-old woman was diagnosed with Fechtner syndrome at 15 weeks gestation. She had a familial history of this disorder; her mother, two siblings and maternal grandmother were also affected. She presented with neither bleeding from the genital tract nor symptoms suggestive of placental abruption. Labor progressed uneventfully and resulted in the birth of a healthy female infant weighing 3436 g at 41 weeks of gestation. The puerperium was uneventful for both mother and infant. ———— Fechtner syndrome; Hereditary thrombocytopenia; leucocyte inclusion body; Alport's syndrome

Fechtner syndrome is a rare autosomal dominant platelet disorder characterized by macrothrombocytopenia, leukocyte inclusions, deafness, nephritis and congenital cataracts. Several cases that resemble Fechtner syndrome have been reported previously in women during pregnancy (Abdel-Fattah et al. 1998). We describe herein the management of a pregnancy complicated with this disorder.

Case Report

A 21-year-old Japanese woman, gravida 2, para 0, was referred to our hospital for antenatal care at 13 weeks of gestation on early March, 1998. She was known to have May-Hegglin anomaly, which had been diagnosed one year earlier by a staff physician at another hospital. Her mother, two siblings and maternal grandmother were also affected. Her mother had chronic nephritis and maternal grandmother had congenital cataracts. The patient had a history of increased tendency to bruise, occasional nosebleeds and blood loss from the gums and ovaries. She had undergone dilatation and curettage twice for induced abortion,

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but had not experienced heavy bleeding.

At 15 weeks of gestation, investigations showed that the platelet count obtained using an automated cell counter was 29×10^9/liter, but the manual platelet count was 65×10^9/liter. The bleeding time was 2 minutes 15 seconds. Giant platelets and light blue inclusion bodies in the cytoplasm of leukocytes were found in the peripheral blood smear that was stained immediately after blood collection. However, the inclusion bodies were only faint in the smear that was stained 6 hours after blood collection. These phenomena were also detected in the peripheral blood smear of her mother, two siblings and maternal grandmother. Therefore, our hematologists performed an electron microscopic examination of the peripheral blood, which demonstrated clusters of fine ribosome-like granules and randomly scattered filaments in the inclusion bodies (Fig. 1). The inclusion bodies also lacked the parallel bundles of filaments characteristic of May-Hegglin anomaly. The above findings confirmed that the patient did not have May-Hegglin anomaly, but Fechtner syndrome.

Antenatal care was undertaken jointly by obstetricians and a consultant hematologist. The patient sufferd no episodes of bleeding from the genital tract or symptoms suggestive of placental abruption during pregnancy. Fetal assessment scans showed normal fetal growth and fetal well being. To select the mode of delivery, fetal blood was sampled at 37 weeks gestation for fetal platelet determination after informed consent was obtained from the patient and her spouse. Blood showed 146×10^9/liter of platelets using an automated cell counter and no inclusion body in leukocytes. Thus, the fetus was confirmed not to be affected by Fechtner syndrome.

Labor was induced at 41 weeks of gestation at the patient's request. Labor

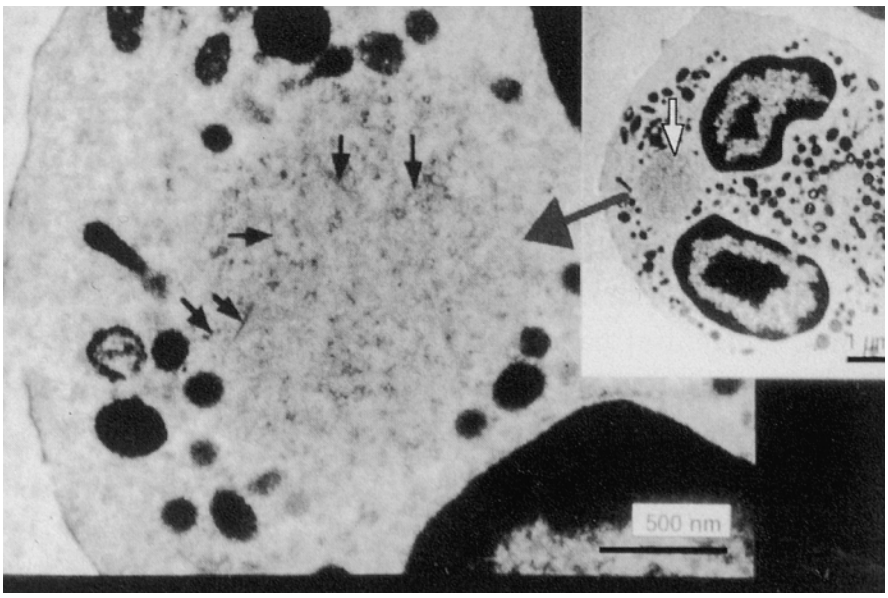

Fig. 1. Neutrophile from the 21-year-old pregnant patient. General morphology is normal. However, small inclusions (white arrow) are present in the cytoplasm. The filaments lie randomly alongside one another (black arrow).

progressed uneventfully and resulted in the birth of a female infant weighing 3436 g, with Apgar scores of 9 and 10 at 1 and 5 minutes, respectively. The placenta was delivered spontaneously under normal conditions and total blood loss was 350 g. The infant's platelet count was 209×10^9/liter. On the third day postpartum, an automated cell counter determined that the maternal platelet count was 73×10^9/liter and the manual platelet count was 108×10^9/liter. The puerperium was uneventful for both the mother and infant.

Discussion

Fechtner syndrome was first described by Peterson et al. (1985) as a variant of Alport syndrome. A few family cases have been reported in Japan. In a review of the obstetric literature, we found no report of Fechtner syndrome during pregnancy. Alport syndrome is characterized by neurosensorial deafness, interstitial nephritis and congenital cataracts. Sebastian syndrome was first described by Greinacher and Eckhardt (1990) and it presents the same hematological changes that occur in Fechtner syndrome without Alport symptoms.

The differential diagnosis between May-Hegglin anomaly, Fechtner syndrome and Sebastian syndrome is made by microscopic or ultramicroscopic differences in the inclusion bodies in leukocytes. The inclusion bodies in Fechtner syndrome and Sebastian syndrome are smaller, located more peripherally in the cell, and stain much more weakly than those in May-Hegglin anomaly in blood smears stained within 4 hours of blood collection. Ultrastructurally, the inclusion bodies in Fechtner syndrome and Sebastian syndrome are not enclosed by a membrane, and consist of a few segments of rough and smooth endoplasmic reticulum, ribosomes and few intermediate filaments. The filaments do not lie in parallel (Greinacher and Eckhardt 1990).

The authors who first described May-Hegglin anomaly cases recommend a vaginal delivery if the qualitative platelet function is normal or corrected by platelet transfusion (Abdel-Fattah et al. 1998). The patient described herein had a normal bleeding time and showed neither bleeding complications nor serious complications at delivery. However, we performed fetal blood sampling by funipuncture to determine fetal affection and platelet count, because there is a lack of information about pregnancies complicated with Fechtner syndrome. Fortunately, the fetus was not affected by this hereditary disease, and the pregnancy and delivery were uneventful. However, further verification is required to confirm the appropriate management of the pregnancy with Fechtner syndrome, especially in patients who have accompanying nephritis.

Reference

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