CASE REPORT

Pregnancy in a patient with Schwachman-Diamond syndrome

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SUMMARY

We present the case of a 26-year-old Caucasian woman with Schwachman-Diamond syndrome with regard to her obstetric and gynaecology requirements, and in particular to her antenatal care across two pregnancies.

BACKGROUND

Schwachman-Diamond syndrome (SDS) is an autosomal recessive disorder that usually presents in infancy with symptoms of pancreatic insufficiency and bone marrow failure including steatorrhoea, growth failure and recurrent infections. It is the second most common inherited exocrine pancreatic dysfunction after cystic fibrosis. Patients may also present with skeletal or hepatic abnormalities.¹

CASE PRESENTATION

The patient was diagnosed with SDS at the age of 4 months, with recurrent bacterial and viral infections (bone marrow failure) and foul nappies (pancreatic insufficiency) on a background of an elder sibling already with the condition. Over the course of her childhood, this was managed with oral pancreatic enzymes and fat-soluble vitamins in increasing doses with every meal and with snacks, and on and off with antibiotics. She engaged well with paediatric and subsequently adult services and demonstrated a reasonable degree of autonomy.

When she reached reproductive age, she appropriately sought advice regarding contraception. Different forms of contraception were discussed, however, the patient was keen to be on oral contraceptives. It was discussed that if her diarrhoea was well controlled with the Creon, then the oral contraceptives should technically be well absorbed, however, the risk of it not working could not be eliminated so she was advised that other methods would be preferred. She also had encounters with the gynaecology services around this time with recurrent Bartholin's cysts and vulval papules.

When she decided that she wanted to start a family, she sought pre-pregnancy counselling. The theoretical risks to the fetus if she developed infections during pregnancy, in particular with parvovirus, were discussed. Risks of pre-eclampsia and intrauterine growth restriction were also considered, given her recurrent infections and pancreatic insufficiency. Also, growth scans would be necessary throughout her pregnancy.

The patient and her partner were advised that they should be referred to a geneticist to establish whether he was a carrier of the disease. If so, there would be a 50% risk that the fetus would inherit

the disorder and a 50% risk of being a carrier but unaffected by the syndrome. If the partner was found to be a carrier, the patient would be offered chorionic villous sampling to establish a prenatal diagnosis.

DIFFERENTIAL DIAGNOSIS

The most common cause of inherited exocrine pancreatic insufficiency is cystic fibrosis and a sweat test should be performed to eliminate this diagnosis. SDS is the second most common. Other causes of bone marrow suppression should also be considered.

TREATMENT

SDS is managed according to symptoms and manifestations, that is, Creon is administered with meals +snacks for pancreatic dysfunction and prophylactic antibiotics for recurrent infections secondary to cyclical neutropenia. Granulocyte-colony stimulation factor can also be considered in cases of severe and persistent neutropenia. On occasion, platelet/blood transfusions may be required to treat deficiencies and rarely a haematopoietic cell transplant should be considered if severe pancytopenia/acute myeloid leucemia is present.

Regular surveillance of blood levels and prevention of infections, for example, by maintaining good oral health, are also required.²

OUTCOME AND FOLLOW-UP

The patient and her partner were soon successful in conceiving and, through the pregnancy, haematology, gastroenterology and obstetric teams monitored the patient regularly. It was discovered that her neutrophils were boosted by the pregnancy and other than a few colds and recurrent urinary tract infections requiring antibiotics, no specific intervention was required. She delivered a healthy baby boy, weighing 7 lbs 5oz, by spontaneous vaginal delivery after a long labour augmented by syntocinon, at 40+12/40.

The patient became pregnant again 2 years later. She required closer monitoring in her second pregnancy because she had additional risk factors.

She was classified as a high risk pregnancy due to:

1. SDS
2. Monochorionic-diamniotic twin pregnancy.
3. Gestational diabetes
4. Family history of venous thromboembolism
5. Asthma

The patient had regular blood tests in pregnancy to monitor her haemoglobin concentration, white cell count and platelets. She had mild neutropenia throughout the pregnancy, but it was never low

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enough to be of concern. Her haemoglobin and platelet count remained normal. She also had regular ultrasound scans for fetal growth.

The mode of delivery was discussed in light of the above risks. An elective Caesarean section was considered, however, a joint decision was made for induction of labour at 36 weeks+3 days, after steroids at 34 weeks to improve fetal lung maturity. The significant risk of an emergency Caesarean section was discussed. She was reviewed by the Anaesthetists and reassured that she could have epidural analgesia if she had a normal platelet count.

The twins were delivered with no complications, the first, weighing 2260 g, by spontaneous vertex delivery, and the second, weighing 2480 g, by assisted breech delivery. Both babies required simple resuscitation as their Apgar scores were 3 at 1 min and 10 at 5 min for the first twin and 3 at 1 min, 10 at 5 min for the second twin.

Postnatally, the patient had a moderate postpartum haemorrhage secondary to uterine atony and was treated for this. She was discharged home on ferrous sulfate, clexane and co-amoxiclav.

None of her children have shown signs of the condition. They did not require genetic testing as her partner was found not to be a carrier of the disease.

DISCUSSION

On studying the literature, only two reports could be found describing a pregnancy in patients with SDS; one described a patient with SDS, Ehlers-Danlos syndrome+urticaria pigmentosa, and reported particularly from the point of view of her bone marrow suppression. Her blood counts were adequate until week 38, when her platelets dropped and she required a Caesarean section.³ The other case had minimal antenatal complications.⁴

Our patient had multiprofessional care by the obstetrician, endocrinologist, anaesthetist and haematologist. She was closely monitored in pregnancy because of her multiple risk factors and the significant risk of needing an emergency Caesarean section. Other than mild neutropenia, the SDS did not cause any problems during pregnancy; however, because it is a rare condition, there was considerable anxiety about her care.

We hope that this case will contribute to the pool of knowledge about the care of patients with SDS in pregnancy.

Learning points

- ▶ Schwachman-Diamond syndrome is a rare autosomal recessive syndrome that typically presents with pancreatic dysfunction and haematological problems.
- ▶ Patients with Schwachman-Diamond syndrome should seek prenatal counselling.
- ▶ During pregnancy they need to be monitored closely by obstetricians experienced in high-risk pregnancies, gastroenterologists and haematologists.

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Competing interests None.

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