

Objective:

Pseudoxanthoma elastium (PXE) is a rare inherited recessive systemic disease of connective tissue. It associates with mutations in the ABCC6 gene (ATP binding cassette subtype C number 6) which at least one ABCC6 mutation is found in about 80% of patients. It is primarily affecting the skin, retina, and cardiovascular system. PXE may have serious complications such as retinal hemorrhages if ocular involvement, also the involvement of the elastic media and intima of the arteries which can lead to intermittent claudication, high blood pressure, GI and cerebral haemorrhage, cardiovascular events such as MI, and angina.

Case report:

A 30-year-old Caucasian lady had 3 normal vaginal deliveries at term with no complication from the Pseudoxanthoma elastium. She has some skin changes in her neck, retinal changes, and intermittent claudication at the time of the diagnosis which was between her second and third pregnancy. The genetic testing is considered due to her signs of the disease and family history of PXE which shows a mutation in the ABCC6 gene. The lady got pregnant with her third child, and she has been followed up in our department. She delivered a healthy infant at 38 weeks by normal vaginal delivery after an uneventful pregnancy. Her follow-up during the pregnancy includes serial growth scan and liaising with other specialties such as cardiology, ophthalmology, and genetics service to reduce the risks of fetal-maternal complications from the disease.

Discussion:

Management of pregnant women with rare diseases has lots of difficulties due to incomplete knowledge and clear guidelines in how to arrange to follow up and manage the pregnancy. From our literature search, while most of the studies and case reports show that pregnancy can be safe in women who have PXE, there are serious maternal complications such as high blood pressure, gastric bleeding, retinal complications and worsening of skin manifestations reported in other studies. In our case, there was no fetal-maternal complication seen related to the Pseudoxanthoma elastium disease with her 3 deliveries.

Conclusions:

Patients with Pseudoxanthoma elastium can have a normal antenatal period followed by normal vaginal delivery if there is no other medical risks impact on their pregnancies. However, antenatal care by a multidisciplinary team (Obstetrician, Ophthalmologist, cardiologist, dermatologist, genetics) may be needed to reduce the risk of disease exacerbation or complications during the pregnancy, labour, and postnatal period. Patients with PXE should not be discouraged from becoming pregnant