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CASE REPORT

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Pseudoxanthoma elasticum and pregnancy

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Abstract A 29-year-old woman affected by pseudoxanthoma elasticum gave birth to her second child in our department, thirteen months after the delivery of her first boy. Her care illustrates many of the potential risks of this rare autosomal systemic disorder. In order to detect any changes due to pregnancy, ophthalmologic and cardiologic screening examinations should be performed in the beginning of the pregnancy and repeated several weeks after the delivery. During labor, epidural anesthesia seems to be more advantageous.

Keywords Pseudoxanthoma elasticum · Pregnancy

Introduction

Pseudoxanthoma elasticum (PXE) is a rare, autosomal systemic disorder characterized clinically by skin, cardiovascular and eye manifestations due mainly to calcification and fragmentation of elastic fibers. Complications during pregnancy have been reported in women affected by PXE. Recently, a 29-year-old woman with this disorder gave birth to her second child in our department, thirteen months after the delivery of her first boy. Her care illustrates many of the classic risks of this disorder.

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Case report

A 28-year-old white woman delivered vaginally a 3,816-g male at 39 weeks following vacuum extraction. She had arrived 6 h earlier in spontaneous labor after spontaneous rupture of the membranes. The Apgar scores were 5, 8 and 9 at 1, 5 and 10 min, respectively. Thirteen months later, she delivered vaginally a 3,670-g female at 39 weeks and 1 day. She had arrived 5 h earlier in spontaneous labor after spontaneous rupture of the membranes. The Apgar scores were 8, 9 and 10 at 1, 5 and 10 min, respectively.

At the age of 22 years, she was diagnosed with PXE by skin biopsy of a skin lesion on the neck. Her younger sister was also affected by the same skin lesions. There was no evidence of consanguinity. DNA analysis has not yet been performed. This was her first pregnancy. She is screened annually by the ophthalmologist; she developed angioid streaks at 26 years. A cardiac echo at the beginning of the first pregnancy was structurally and physiologically normal with an ejection fraction of 60%.

The course of the pregnancies was uneventful apart from an increase in the skin manifestations in the neck. During her first pregnancy, a biochemical screening (triple test) for trisomy 21 at 17 weeks and an ultrasound examination at 23 weeks were interpreted as normal. At 32 weeks, an ultrasound revealed a placenta with more areas of calcification than expected. However, the umbilical and uterine Doppler flow velocities remained normal. Puerperium recovery and healing of the episiotomy wound were uneventful. The placenta showed normal morphological features apart from pronounced calcifications on the maternal side. Histological examination confirmed the presence of calcifications crossing the whole thickness from one side of the placenta to the other.

During the second pregnancy, she had an amniocentesis at 18 weeks because of an increased risk for trisomy 21 during the second trimester biochemical

screening. The caryotype was 46XX. The rest of this pregnancy was similar to the first one.

Cardiologic and ophthalmologic screening examinations were repeated 10 weeks after each delivery; there were no changes from baseline.

Discussion

Pseudoxanthoma elasticum is an autosomal dominant/recessive disorder with variable expression and penetrance. It has an estimated prevalence of 1:70,000 to 1:100,000 live births and is characterized by multiple systemic manifestations including skin lesions, and abnormalities of the Burch's membrane in the eye and the cardiovascular system. The abnormalities share progressive mineralization of elastic fibers and abnormal deposition of aggregates of matrix components within the dermis and in soft tissues.

The most common presentation is of skin papules that are ivory- to yellowish-colored, raised and vary in size from 1–3 mm. The papules may have a linear or reticular arrangement and coalesce into plaques. Though characteristic, there are other disorders that may present similarly, such as focal dermal elastosis, beta-thalassemia, and the Paget's disease or osteoectasia [3]. In many instances, the skin becomes wrinkled and redundant, hanging in folds. These folds may become more prominent during pregnancy. On average, the diagnosis of PXE is made at 22 years, just as the patient described.

Ocular signs eventually develop in most patients with PXE. The usual sequence of abnormalities is mottled hyperpigmentation of the retina, angioid streaks, peripapillary atrophy with or without white glial tissue formation, and finally, subretinal neovascularization. The latter often leads to a disciform scar in the macula, producing decreased visual acuity [3].

Cardiovascular complications in PXE are relatively frequent, but exact prevalence is unknown. Cardiovascular signs and symptoms include angina pectoris, diminished pulse waves, hypertension, restrictive cardiomyopathy, mitral valve prolapse and stenosis, fibrous thickening of the endocardium and atrioventricular valves, and sudden death at younger age [3]. Apart from the latter, the most serious complication of PXE is accelerated arteriosclerosis.

Given the clinical heterogeneity and apparent multiple mechanisms of inheritance, it is difficult to both diagnose PXE accurately and calculate correctly the genetic risks for counseling purposes. The PXE gene has been localized to chromosome 16p13.1. The results of linkage studies, which suggest allelic heterogeneity in a single gene disease, could account for both autosomal recessive and autosomal dominant forms of PXE. The PXE gene encodes the ABCC6, a protein belonging to the ABC transport cassette system. To date, the physiological function and the involvement of ABCC6 in the PXE phenotype remain unclear [3].

There is great potential risk for both mother and fetus during pregnancy. Apart from a slight increase in the spontaneous first trimester abortion rate, patients with PXE had normal pregnancies, labors and deliveries. However, cardiac arrhythmia, congestive heart failure, hypertension, intrauterine growth restriction and thromboembolic phenomena have also been reported [1, 2]. Gastrointestinal hemorrhage too is reported during pregnancy, and aspirin and non-steroidal anti-inflammatory drugs should be avoided.

Both gross and light microscopic examinations failed to reveal dramatic differences between placentas of PXE women and controls in terms of size, infarcts, thrombi, inflammatory lesions or vessels [2]. However, these investigators noted that mineralization was more pronounced in the placentas of women affected by PXE.

During labor, an epidural has the advantage of excellent pain relief and the avoidance of general anesthesia. Since women with PXE may have accelerated atherosclerosis, it is important to maintain blood pressure and avoid tachycardia during labor. Patients with PXE may develop retinal bleeding during the second stage of labor, and consideration should be given to shortening it through the judicious use of either forceps or vacuum [1].

In conclusion, this case illustrates the factors that have to be taken into consideration during the pregnancy of a PXE patient. In order to detect any changes due to pregnancy, it is important to have ophthalmologic and cardiologic screening examinations early in the pregnancy and several weeks after the delivery. Although there is a great potential risk for both mother and fetus during the pregnancy, this case shows the contrary.

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