CASE REPORT

Pregnancy with Ehlers Danlos Syndrome Type II

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Abstract

The occurrence of pregnancy in females with Ehlers Danlos syndrome is not common. This is a rare inherited connective tissue disorder that has been associated with numerous pregnancy complications ranging from bruising to maternal death. More than 10 variants of the Ehlers Danlos syndrome have been identified based upon either biochemical or clinical findings; and, different major and minor discriminatory criteria have been defined for each. Type I and type II variants of the classical Ehlers Danlos syndrome are well known, with type II being the less severe one. Despite constituting 35 percent of all cases, pregnancy associated with type II is rarely reported, due to the rare observation of complications. The case of pregnancy of a 29 year old, second gravida with Ehlers Danlos syndrome (Beighton score 5/9) is reported here. The patient developed non-immune hydrops and intrauterine fetal death at 34 weeks pregnancy. Her previous delivery was by cesarean section.

Keywords: Ehlers Danlos syndrome, pregnancy, vaginal birth after caesarean

Özet

Tip II Ehlers Danlos Sendromunda Gebelik

Ehlers Danlos sendromu olan kadın hastalarda gebelik sık görülmez. Bu, nadir görülen kalıtımsal bir bağ dokusu hastalığı olup, hafif ekimozlardan anne ölümüne kadar değişen pek çok gebelik komplikasyonu ile klinik verebilir. Bugüne kadar biyokimyasal ya da klinik bulgulara göre 10 kadar Ehlers Danlos sendromu tipi tarif edilmiştir. Her bir tip için değişen majör ve minör tanı kriterleri tanımlanmıştır. Ehlers Danlos sendromu tip I ve II varyasyonları iyi bilinmektedir ve tip II formu daha hafif seyirlidir. Tüm Ehlers Danlos sendromu olgularının %35'ini kapsasa da tip II formunda gebelik, komplikasyon riskinin düşük olması nedeniyle nadiren bildirilmiştir. Bu yazıda, ikinci gebeliğinde Ehlers Danlos sendromu olan 29 yaşında bir olgu bildirilmektedir (Beighton skoru 5/9). Gebeliği döneminde hastada nonimmün hidrops gelişmiş ve 34. haftada fetüs *in utero* ölmüştür. Hastanın önceki doğumu sezaryen ile yaptırılmıştı.

Anahtar sözcükler: Ehlers Danlos sendromu, gebelik, sezaryen sonrası vajinal doğum

Introduction

Occurrence of pregnancy with Ehlers Danlos syndrome is not common. This is a rare inherited connective tissue disorder that has been associated with numerous pregnancy complications ranging from bruising to maternal death. More than 10 variants of the Ehlers Danlos syndrome have been identified based upon either biochemical or clinical findings (1). Different major and minor criteria have been defined for each type. Type I and type II are the better known variants of the classical Ehlers Danlos syndrome, with type II being a less severe one (2,3). Pregnancy in association with type II is rarely reported, despite the fact that it constitutes 35 percent of all cases (4). This is due to the milder presentation withrare occurrence of complications. Here, we are reporting the case of a type II Ehlers Danlos syndrome patient. Whose pregnancy was complicated by recurrent non-immune hydrops and the rupture, after vaginal delivery, of uterine scar of the previous cesarian section.

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Case Report

A 29 years old gravida 2, para 1 was referred at 31 weeks of gestation with fetal hydrops. Fetal ascites, scalp edema and pleural effusion were seen by ultrasonography. The patient had a history of chronic arthralgia and myalgia since the age of 18 years. She had the characteristic facial appearance including a thin pinched nose and large prominent eyes with hollow cheeks. Her hands and feet were large, relative to her body stature. The skin of her face, hands and feet appeared prematurely wrinkled and aged. It was excessively soft and strechable although the vasculature could not be seen through it. There was history of easy bruisability. Excessive laxity of the metacarpophalangeal and interphalangeal joints of hands and feet was confirmed on physical examination. Her Beighton score was 5/9 (5). She had an unintelligible speech and the absence of both the inferior labial frenulum and lingual frenulum, which are characteristic of the Ehlers Danlos syndrome further confirmed the clinical diagnosis.

In her first pregnancy, four years back, she developed polyhydramnios at 28 weeks of gestation for which no investigations were carried out. An elective cesarean was performed at 35 weeks in view of the increasing maternal discomfort, the records of which were not available. A 2500 gram male infant with Apgar score of 7,8 was delivered. The baby was hydropic at birth, developed neonatal jaundice within 48 hours and received phototherapy. There was no evidence of neonatal sepsis. On follow up, the child had learning disability.

The patient was normotensive, non-diabetic with 0 positive blood group. Blood tests for syphilis, toxoplasmosis and herpes were negative. Although ultrasonography and an echocardiogram revealed no structural fetal anomalies, there was severe intrauterine growth restriction. At 32 weeks of the pregnancy Doppler showed absent end-diastolic flow in umbilical artery leading to intrauterine fetal demise at 34 weeks. A weekly coagulogram was done, awaiting spontaneous labor to occur. The patient insisted on vaginal delivery. At 39 weeks a cervical assessment was done. As the Bishop's score was unfavorable, a 16F Foleys catheter was inserted intracervically as a ripening agent. She went into labor within 4 hours and did not require any oxytocin augmentation. She delivered a macerated baby of 1000 grams after 6 hours of labor. Autopsy of the baby did not reveal any malformations. Immediately following delivery of the placenta, the patient started bleeding heavily. A per vaginum examination revealed a cervical tear going upto the fornix. She was taken for immediate laparotomy suspecting scar rupture. At laparotomy under general anaesthesia, there was a dehiscence of 4 cm of previous uterine scar on the right side, which was extending down to the cervix which had to be repaired. There was excessive oozing from the skin incision, which was controlled with difficulty by multiple mattress sutures. The wound healed without any complications. The patient was discharged satisfactorily after 15 days of stay in the hospital.

Discussion

The incidence of Ehlers Danlos syndrome in pregnancy has been estimated at 1 in 150 000. The major features characterizing the classical form is the presence of hyperextensible skin, hypermobile joints and wide thin scars. The minor criteria include presence of smooth velvety skin, muscle hypotonia, easy bruising and a positive family history. Types I-III are the most common, each representing approximately 30% of reported Ehlers Danlos syndrome cases. Type IV is found in 10% of cases, with the remaining types being exceptionally rare. Not all types of Ehlers Danlos syndrome are necessarily associated with unacceptable morbidity or mortality rates (6). There is however a paucity of data from large studies on which to base firm conclusions about the risks of pregnancy in association with various forms of this syndrome. Gravis' type I Ehlers Danlos syndrome is associated with tissue fragility, hemorrhage and poor wound healing. Maternal mortality with "ecchymotic" type IV Ehlers Danlos syndrome is quoted to be as high as 25% because of arterial rupture. Since type I and type IV are most severe forms, some authors have suggested that perhaps pregnancy should be avoided in these patients (7,8). Reports on type II Ehlers Danlos syndrome patients describe a relatively benign obstetric outcome with maternal complications arising primarily from tissue laxity in limb mobility. Preterm premature rupture of membranes has been reported to be very common in these patients (9).

However, our patient's course was complicated by fetal hydrops of unknown etiology, intrauterine fetal death and the rupture of uterine scar of the previous cesarian section following vaginal birth. She did not receive any oxytocics, had a rapid progression of labor and scar rupture occurred in the second stage of labor, which could not have been prevented.

It appears that there might be a defective healing process in women with Ehlers Danlos syndrome type II. An electiv cesarean may be considered in those with previous cesarean.

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