Case Report

A 29-year-old woman was referred for pre-pregnancy counseling for classic-type Ehlers Danlos syndrome with joint hypermobility, recurrent dislocations, hyperextensible skin, keloid scarring and fatigue. She was advised of a 1:2 chance of having an affected child. The patient conceived spontaneously. At 11 weeks she ceased work due to significant joint discomfort. Increasing gravity caused a marked worsening in her symptoms of lower back and pelvic pain. At 29 weeks she was reviewed by a psychiatrist for daily panic attacks triggered by fear of premature labour, musculoskeletal injury and worsening pain. At 33w6d she presented in preterm labour. Steroids were administered and a baby boy at 2520g delivered by uncomplicated caesarean section.

She presented eleven months later, early in her second pregnancy. In view of the previous pregnancy, early steroid administration, screening of cervical length during the third trimester and elective caesarean section at 38 weeks were planned. The patient again suffered with severe back, pelvic and joint pain, and anxiety symptoms. She was admitted with threatened preterm labour at 25, 26, 28 and 32 weeks gestation and was treated with nifedipine and betamethasone. Daily painful contractions, debilitating joint pain and pubic symphysis diastasis occurred. Due to intractable pain, caesarean section was performed at 34w0d and a baby girl at 2395g was delivered. No abnormal scarring of the caesarean section wound has occurred. The joint pain improved slightly postpartum but has not returned to pre-pregnancy levels. Both children (aged 2 and 3) have been diagnosed with hypermobility EDS.

References

2. Symons S, Sym D, Maitland P. Comprehensive molecular analysis demonstrates type V collagen mutations in over 90% of patients with classic EDS and allows to refine diagnostic criteria. Hum Mutat. 2012;33:1485.

Complications of Classic Ehlers Danlos Syndrome in Pregnancy

V Watson, Logan Hospital

Ehlers-Danlos syndrome encompasses a group of genetic disorders of collagen synthesis with an incidence between 1 in 5000 and 1 in 20000(1). Classic Ehlers-Danlos syndrome (EDS) results from autosomal dominant gene mutations causing large and small joint hypermobility, hyperextensible “doughy” skin and abnormal or atrophic wound healing(2). Data from rare case reports indicate mothers are at increased risk of pelvic pain and instability, cervical incompetence, preterm prelabour rupture of membranes, uterine rupture, complicated pelvic floor trauma, uterine prolapse, poor wound healing and post partum haemorrhage(3). In addition chronic dyspareunia secondary to symphysis pubis diastasis, bladder and bowel prolapse are more common.

This case demonstrates a propensity for mothers with classic Ehlers-Danlos syndrome to experience preterm labour and debilitating pain related to joint laxity. Close surveillance (including cervical length), psychosocial support and possibly early antenatal steroid administration are indicated.

Important Points

• Pre-pregnancy counselling is important including consideration of pre-implantation genetic diagnosis, chorionic villi sampling or amniocentesis
• There is no clear advantage to vaginal or caesarean delivery

Discussion

Figures 1 and 2: Physical signs of joint hypermobility

Ethics Approval: Written consent was obtained from the patient.