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Ehlers-Danlos Syndrome In Pregnancy: A Case Report Of Arthrochalasia Type 2 With Successful Outcome.

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Abstract:

Background : Ehlers-Danlos Syndrome (EDS) is a heterogeneous group of inherited connective tissue disorders, characterized by joint hypermobility, skin hyperextensibility, and tissue fragility. Though most women with hypermobile EDS experience uncomplicated pregnancies, awareness of potential complications such as recurrent joint dislocations, autonomic pain, and postural orthostatic tachycardia syndrome , Preterm Labour , Postpartum haemorrhage , Complicated perineal lacerations are crucial. Early diagnosis and subtype characterization aid in pre-pregnancy counselling and help manage obstetric and anaesthetic risks.

Case Presentation : A 27-year-old primigravida with EDS arthrochalasia type 2 presented with joint hypermobility, skin hyperextensibility, and fragility. Genetic testing confirmed the diagnosis. The pregnancy was largely uneventful, but she experienced pelvic girdle pain. At 36 weeks of gestation, she underwent an emergency caesarean section due to maternal request. No complications were noted intra-operatively or post-operatively, and both mother and baby had a good outcome.

Conclusion: Increased awareness of EDS during pregnancy, coupled with a multi-disciplinary approach, can result in favorable maternal and fetal outcomes. Early diagnosis and tailored care are essential in minimizing obstetric and anaesthetic risks.

Index Terms : Ehlers-Danlos Syndrome, Pregnancy, Joint Hypermobility, Arthrochalasia Type 2, Obstetric Complications, Cesarean Section

Introduction:

Ehlers-Danlos Syndromes (EDS) are a group of rare, inherited connective tissue disorders characterized by joint hypermobility, skin hyperextensibility, and tissue fragility. The various types of EDS have different genetic causes, clinical presentations, and inheritance patterns. The incidence of all types of EDS in pregnancy is relatively low, ranging from 1 in 5,000 to 1 in 20,000.[1] However, given the heterogeneity of EDS, it's important to understand the specific subtype in order to provide optimal care.

Among the different types of EDS, the classical type accounts for approximately 30% of cases, the hypermobility type represents around 35%, and vascular EDS accounts for 10-30%. While the hypermobility type is the most common, women with this subtype typically have uncomplicated pregnancies. However, certain risks and complications are more prevalent and should be closely monitored.[2]

Potential Complications in Pregnancy:

1. Recurrent Joint Dislocations: Due to the hypermobility of the joints, pregnant women with EDS may experience dislocations, which could affect mobility and cause significant discomfort.[2]
2. History of Surgical Joint Stabilization: Women with prior joint stabilization surgeries may face challenges during labor or experience joint instability due to pregnancy-related changes in ligamentous laxity.[2]
3. Autonomic Pain and Postural Orthostatic Tachycardia Syndrome (POTS): These conditions, which are common in hypermobile EDS, can result in significant discomfort and complications during pregnancy.[3]
4. Preterm Labour: The fragility of connective tissues can contribute to premature rupture of membranes and early labour.[1]
5. Postpartum Hemorrhage: Due to vascular fragility, there is an increased risk of postpartum bleeding and complications related to wound healing.[5]
6. Complicated Perineal Laceration: The skin and connective tissue's fragility can lead to more severe tearing during delivery.[4]
7. Uterine Rupture: The increased tissue fragility in the uterine wall, especially in vascular EDS, increases the risk of uterine rupture during pregnancy or labour.[3]
8. Poor Wound Healing: A common feature of EDS is delayed or impaired wound healing, which can lead to complications after cesarean sections, episiotomies, or any surgical procedures during pregnancy .[2]

Early diagnosis of EDS and accurate classification of the specific subtype is essential in pre-pregnancy counseling, antenatal care planning, and risk assessment for obstetric complications. A thorough understanding of the specific subtype allows for tailored management to minimize risks during pregnancy, labor, and the postpartum period.Given the complexities of managing a pregnancy in a woman with EDS, a multidisciplinary approach is key to optimizing outcomes. The involvement of obstetricians, geneticists, anesthesiologists, physiotherapists, and pain management specialists can help address the various aspects of care. This ensures that both the mother and baby receive appropriate support and intervention, from monitoring joint instability to managing potential cardiovascular and wound healing challenges.

CasePresentation :

A 27-year-old primigravida presented to the antenatal clinic for her first visit. On physical examination, she exhibited skin hyperextensibility, joint hypermobility, and skin fragility. The skin had a velvety texture, was highly stretchable, and demonstrated hyperextensibility. The joints were lax and hypermobile, though there was no history of joint dislocation. The patient had normal intelligence and no neurological deficits. Examination of other systems was unremarkable.

Basic blood investigations were normal. She conceived after two cycles of ovulation induction, with no relevant family history of similar features. Given her clinical presentation, genetic testing was advised, and Whole Exon Sequencing revealed the diagnosis of Ehlers-Danlos Syndrome, Arthrochalasia Type 2 (OMIM#61721), with a gene involvement of *COL1A2*.

In the first trimester, the patient underwent a nuchal translucency (NT) scan and double marker test, both of which showed low risk. An electrocardiogram (ECG) and 2D echocardiogram were normal. She reported persistent pelvic girdle pain throughout the pregnancy, which was managed conservatively. Throughout her pregnancy, her pulse rate ranged between 110-120 beats per minute.

The second trimester was uneventful, with an anomaly scan revealing no abnormalities. An interval growth scan at 30 weeks also showed normal fetal growth. At 34 weeks, steroid prophylaxis was administered in preparation for a possible preterm delivery. At 36 weeks gestational age, the patient presented in preterm labor and underwent an emergency lower segment cesarean section (LSCS)due to maternal request(CDMR).Lower caesarean segment section was carried out with general anaesthesia with full preparation of potential intrapartum complications.Abdomen was opened with pfannensteil incision , uterus opened with Kerr's ncision.Tissues were markedly thin and fragile and rectus sheath was noted to be papery thin . There were no intra-operative complications, and a live male infant weighing 2.7 kg was delivered. Both intra-operative and post-operative bleeding were within normal limits, and wound healing was uneventful. Post-operative antibiotics were given for 5 days.Patient was discharged on day 3 post-operative and asked to visit again on day 7 for evaluation of wound healing.On Postnatal day -7 , baby also evaluated for same variant , A heterozygous missense variant in exon 45 of the COL1A2 gene was detected in the son. Wound healing was uncomplicated. The puerperium was uneventful.

Discussion:

Ehlers-Danlos Syndrome (EDS) is a connective tissue disorder that can present with varying degrees of severity. In the case of hypermobile EDS (hEDS), pregnancy may proceed without significant complications in most cases. However, awareness of the potential risks is essential for proper management. The presence of hypermobile joints, skin fragility, and increased skin extensibility can complicate labor and delivery, particularly in regard to anaesthesia, wound healing ,and surgical procedures.

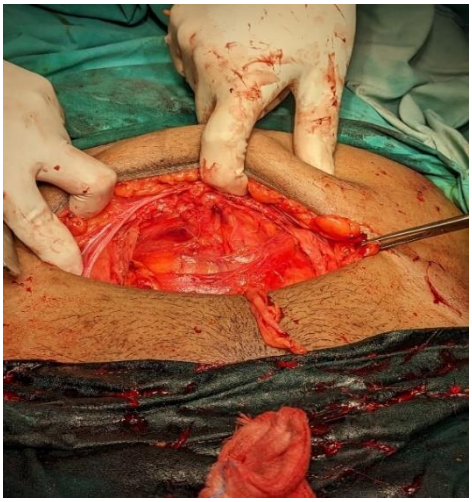
This case highlights the importance of early diagnosis and multi-disciplinary management in ensuring a favourable outcome for both the mother and the baby. Although this patient had a relatively uncomplicated pregnancy with the exception of pelvic girdle pain, the risk of joint dislocations and other musculoskeletal complications could have posed challenges in her care. Additionally, the thinness of the rectus sheath noted during the cesarean section underscores the increased risk of tissue fragility in these patients.

Several studies have emphasized the need for careful pre-pregnancy counselling for women with EDS, particularly in relation to the risk of obstetric complications, including preterm labour and the need for cesarean delivery. A proactive approach to care, including genetic counselling, regular antenatal monitoring, and a coordinated care plan, can lead to better outcomes.

The role of genetic testing in identifying specific subtypes of EDS is critical, as it helps guide management strategies and tailor care to the individual patient's needs.

Hypermobile knee joint

Hypermobile joints in hand



Papery thin rectus sheath

Conclusion:

Ehlers-Danlos Syndrome in pregnancy, though rare, requires careful management to ensure a positive outcome. Early recognition, subtype classification, and a multi-disciplinary approach to care are essential in preventing complications and optimizing both maternal and fetal health. This case illustrates the importance of comprehensive care for women with EDS during pregnancy, with favourable outcomes possible through appropriate management.

References:

1. Malfait, F., et al. (2017). *The Ehlers-Danlos syndromes: a comprehensive review of clinical and genetic features*. Genetics in Medicine, 19(5), 1500-1507. https://doi.org/10.1038/gim.2017.39
2. Tinkle, B., et al. (2017). *The hypermobility type of Ehlers-Danlos syndrome: clinical features and management strategies*. Rheumatology, 56(10), 1766-1774. https://doi.org/10.1093/rheumatology/kew477
3. Byers, P. H., et al. (2017). *Ehlers-Danlos syndrome: diagnosis and management*. American Journal of Medical Genetics Part C: Seminars in Medical Genetics, 175(1), 4-17. https://doi.org/10.1002/ajmg.c.31522
4. Castori, M., et al. (2017). *Ehlers-Danlos syndrome: pathogenesis, diagnosis, and management*. American Journal of Medical Genetics Part C: Seminars in Medical Genetics, 175(1), 4-17. https://doi.org/10.1002/ajmg.c.31522
5. De Witte, K., et al. (2016). *Pregnancy and delivery in women with Ehlers-Danlos syndrome: a case series*. American Journal of Obstetrics and Gynecology, 215(5), 626-632. https://doi.org/10.1016/j.ajog.2016.02.017