Reduction mammaplasty in an adolescent with Ehlers-Danlos syndrome: a case report

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Background: Hypermobile Ehlers-Danlos syndrome (hEDS) is a connective tissue disorder that results in joint hypermobility and tissue fragility. Symptoms of macromastia may be exacerbated in patients with concomitant hEDS. Surgeons are often hesitant to operate on patients with hEDS given their increased propensity to develop surgical complications, including wound dehiscence, severe bruising, and hematomas, but there is a paucity of literature actually describing surgical complications in these patients. This is further limited in the pediatric and adolescent populations. To our knowledge, this is the first report of breast reduction in an adolescent patient with any form of Ehlers-Danlos syndrome (EDS).

Case Description: This case report describes an adolescent patient with hEDS who underwent bilateral breast reduction for symptomatic macromastia with an uncomplicated recovery. An 18-year-old female presented to the plastic surgery clinic with chronic back, neck, and arm pain, poor body image and low self-esteem, inability to participate in sports and social activities due to emotional distress and discomfort, poor posture, difficulty fitting into clothes, and inframammary intertrigo all related to large breast size. She previously failed physical therapy and other forms of conservative management. On physical examination, she had large, ptotic, heavy dense breasts with significant striae. The areola size was 7 cm on the left and 7.5 cm on the right, the breast footprint base was 13 cm on the left and 14 cm on the right, the nipple to inframammary fold distance was 11.5 cm on the left and 11.5 cm on the right, and the suprasternal notch to nipple distance was 30 cm on the left and 30.5 cm on the right. She was diagnosed with macromastia and underwent bilateral mammoplasty with Wise-pattern and inferior pedicles. The patient recovered uneventfully after the procedure and followed up postoperatively with both the physical medicine and rehabilitation and plastic surgery departments. She subjectively reported a reduction in neck and back pain.

Conclusions: We propose that hypermobile EDS may not be a contraindication to surgery in adolescents and acceptable outcomes can be achieved.

Keywords: Breast reduction surgery; Ehlers-Danlos syndrome (EDS); adolescent; case report; hypermobility

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Introduction

Ehlers-Danlos syndrome (EDS) is a group of genetic connective tissue disorders involving mutations in collagen biosynthesis, processing, or folding, the extracellular matrix structure and function, the glycosaminoglycan biosynthesis pathway, or intracellular processing (1). The disease is characterized by joint hypermobility, poor wound healing, vascular and tissue fragility, and skin hyperextensibility, predisposing patients to chronic joint pain. Hypermobile EDS (hEDS), previously known as type III EDS, is the most common form (1). These patients commonly have joint pain and muscle pain that may be exacerbated by macromastia. An argument can be made that these patients may experience greater benefit from a reduction mammoplasty than patients without hEDS. However, surgeons may be hesitant to operate on these patients given their complex diagnosis and increased propensity to develop surgical complications, including wound dehiscence, severe bruising, and increased risk of hemorrhage and hematomas (2-7). Due to the fragile connective tissue and blood vessels, delayed wound healing, and chronic pain, patients with EDS may have less favorable surgical outcomes compared to the general population (8-10).

Most of the published studies focus on surgery in adults with EDS (3,10-12). However, there are few published studies describing cases of plastic surgery in patients with EDS (2,4,6,13). There are even fewer studies describing surgery in pediatric and adolescent patients (5,14,15). In many of these case studies, patients' EDS subtype was not known or stated. Each subtype is due to a unique mutation leading to a distinct set of symptoms and potential surgical complications. Moreover, classification of EDS subtypes has changed multiple times over the last several years. Therefore, previous studies broadly describing surgical complications of EDS are not necessarily valid in informing a surgeon of the potential complications their patient may encounter and cannot be universally applied to hEDS patients or patients with mild manifestations of the disease.

This article discusses an adolescent patient (defined by the World Health Organization as ages 10–19 years old) with hEDS and macromastia who underwent bilateral reduction mammoplasty for symptomatic macromastia with uncomplicated healing and reduction in symptoms. We propose that hEDS is not an absolute contraindication for surgery and that patients with hEDS show improvement of symptoms regarding joint and muscle pain. To our knowledge, this is the first report of breast reduction in an adolescent patient with any form of EDS. We present the following case in accordance with the CARE reporting checklist (available at https://amj.amegroups.com/article/view/10.21037/amj-22-61/rc).

Highlight box

Key findings

- Hypermobile Ehlers-Danlos syndrome may not be a contraindication to surgery in adolescents and acceptable outcomes can be achieved.

What is known and what is new?

- There has traditionally been concern for post-surgical healing complications in patients with hypermobile Ehlers-Danlos syndrome. There is a paucity of literature surrounding surgical wound healing in patients with hypermobile Ehlers-Danlos syndrome, and this is further limited in the pediatric and adolescent population.
- This case report describes an adolescent patient with hypermobile Ehlers-Danlos syndrome who underwent bilateral breast reduction for symptomatic macromastia with an uncomplicated recovery.

What is the implication, and what should change now?

- A diagnosis of hEDS should not be considered an absolute contraindication to reduction mammoplasty in patients with macromastia and careful consideration should be given to this patient population when deciding whether to offer them surgery.

Case presentation

All procedures performed in this study were in accordance with the ethical standards of the institutional and/or national research committee(s) and with the Helsinki Declaration (as revised in 2013). Written informed consent was obtained from the patient for publication of this case report and accompanying images. A copy of the written consent is available for review by the editorial office of this journal. An 18-year-old female presented to the plastic surgery department with macromastia causing chronic back, neck, and arm pain, poor body image and low self-esteem, inability to participate in sports and social activities due to emotional distress and discomfort, poor posture, difficulty fitting into clothes, and inframammary intertrigo. Her breast development started a year before her menstrual cycle, and her cup size grew from an "A" to a "F" in one year. At the time of evaluation, she wore a 38G. Her breast size was stable and fluctuated minimally with weight change for the year prior to evaluation.

Her medical history was significant for mild spastic

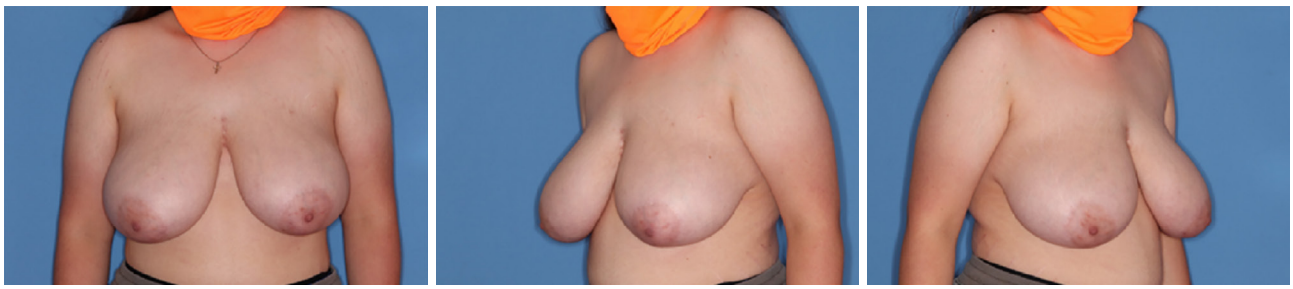

Figure 1 Anterior-posterior view, left anterior oblique view, and right anterior oblique view of the patient's preoperative breasts.

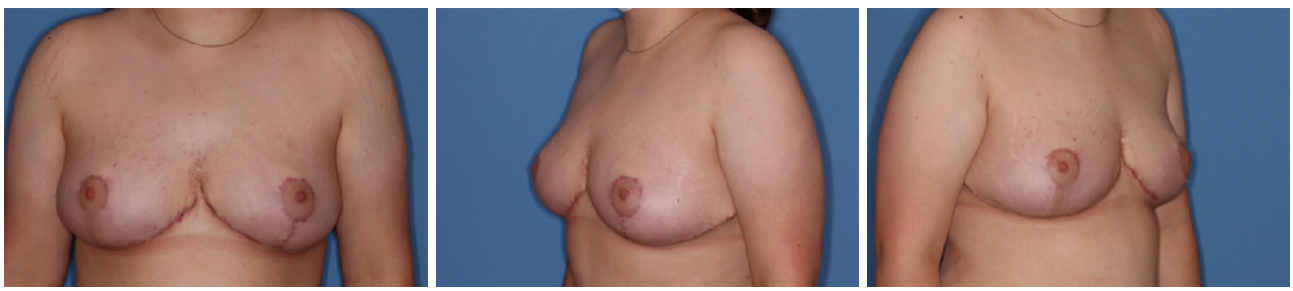

Figure 2 Anterior-posterior view, left anterior oblique view, and right anterior oblique view of the patient's postoperative breasts 3 months after surgery.

cerebral palsy in her ankles, anxiety, and Duane syndrome Type II. For a year and half, she had been followed by the hospital's Complex Pain Clinic, including physical therapy, but conservative therapies were unsuccessful in addressing her symptoms. She was diagnosed with hEDS by a geneticist and was followed in the hypermobility clinic.

On physical examination, she had large, ptotic, heavy dense breasts with significant striae (*Figure 1*). The areola size was 7 cm on the left and 7.5 cm on the right, the breast footprint base was 13 cm on the left and 14 cm on the right, the nipple to inframammary fold distance was 11.5 cm on the left and 11.5 cm on the right, and the suprasternal notch to nipple distance was 30 cm on the left and 30.5 cm on the right. After meeting with the plastic surgeon, she underwent evaluation by the breast psychologist for surgical readiness and preparation. Surgical treatment options were discussed with the patient and her family. Based on her workup, she was deemed appropriate for a bilateral reduction mammoplasty. The patient and family were made aware of potential surgical complications related to hEDS. Bilateral mammoplasty with Wise-pattern and inferior pedicles was performed. The senior author resected 1,011 g of tissue from the right breast and 971 g of tissue from the left breast. A 2-layer closure was performed using deep dermal 3-0 PDS and 3-0 Monocryl along the inframammary fold and vertical incision. Around the nipple areolar complex, 3-0 and 4-0 Monocryl were used to close the deep dermal layer and a 4-0 Monocryl running subcuticular suture was used to secure the epidermal edges. No surgical drains were used. Dermabond and Steri-Strips were applied to the incisions, and she was discharged home on the day of surgery. Steri-Strips were left on for 2 weeks. The patient was also initially dressed in a soft sports bra, which she wore for 6 weeks unless showering. The patient was advised to keep the incisions out of the sun and massage any raised incision lines. This patient applied silicone sheeting to the scars as they developed and was advised to continue to do this for 1 year post-op.

The patient attended appointments with the physical medicine and rehabilitation department for one year after the surgery and followed up with the plastic surgery department for 3 months postoperatively. Surgical pathology revealed pseudoangiomatous stromal hyperplasia with focal fibroadenomatous-like changes. The patient recovered uneventfully after the procedure, with no evidence of wound dehiscence and no complications including hematoma, seroma, pathologic scar formation, or scar widening in the initial postoperative period (*Figure 2*). She subjectively reported a significant reduction in neck and back pain and

her nipple sensitivity was intact but slightly reduced. Typical follow-up courses for breast reductions are as follows: 1–2 weeks, 4–6 weeks, 3 months and 1 year. Patients with EDS are seen on the same postoperative timeline. Any patient with wound healing complications is seen more frequently. In our experience, this has not been the case for hEDS patients.

Discussion

Although considered to be the least severe type of EDS, hEDS represents 80–90% of cases of EDS and has a prevalence in the general population of 1/5,000 (2). Given the variability in symptoms, the actual prevalence may be under reported.

Compared to the other types of EDS, hEDS poses less surgical risk due to less fragility of the vasculature, skin, and soft tissue (7). Most of the case reports and studies cited in this manuscript do not specify which EDS subtypes their patients had; and if they do, most discuss severe complications as a result of more debilitating forms of EDS. Nonetheless, there is increased risk for surgical complications in hEDS compared to the average patient (7). Children with EDS who undergo surgery are at greater risk for friable tissues and hematomas (5). However, successful surgical outcomes in patients with hEDS can be achieved with careful anesthesia and perioperative management (7).

Macromastia is associated with physical and psychological symptoms that lead to poor quality of life compared to the normal population (16). Symptoms may include low self-esteem, intertrigo, difficulty finding clothes that fit, difficulty exercising, shoulder grooving, poor posture, and neck, back, shoulder, and breast pain (16,17). Bilateral reduction mammoplasty has been shown to reduce pain and improve quality of life (16,17). Wampler *et al.* found that mean preoperative scores for satisfaction with breast, psychosocial well-being, and sexual well-being were below the normative scores, but postoperative scores were above the normative scores (18). Teenagers who underwent reduction mammaplasty were able to achieve sustained relief of symptoms of back, neck, and shoulder-strap pain (19). Overall, breast reduction surgery in adolescents offers significant benefits that outweigh the low risk of regrowth of excess breast tissue and slightly diminished breastfeeding capabilities (20).

In this case report, we discussed an 18-year-old female with macromastia and hEDS who underwent bilateral reduction mammoplasty. Surgical complications for patients with hEDS may consist of wound dehiscence, hypertrophic scarring, and hematomas, making surgeons reluctant to operate (5,8-10). However, she experienced no postoperative complications, and her preoperative symptoms improved. To our knowledge, this is the first description of an adolescent with hEDS who received breast reduction surgery. We propose that reduction mammoplasty in adolescents with hEDS may not be contraindicated, but instead may offer substantial improvement of the physical and psychological symptoms associated with macromastia. Furthermore, hEDS patients may benefit more than the general population from surgical management because the features of their disease leave them vulnerable to more severe manifestations of macromastia.

Our study has several limitations. It discusses successful breast reduction surgery in a single patient, which cannot be easily applied to a greater population. Follow-up for this patient was 1 year with physical medicine and rehabilitation (only 3 months in the plastic surgery clinic), but long-term follow-up may be needed to determine whether the operation resulted in sustained relief of symptoms. This patient had significant benefit from her reduction mammoplasty but we recognize that not all patients may experience the same relief. It is often difficult to discern if symptoms of pain are related to the weight of the breasts or the other underlying diagnosis, such as hEDS joint pain, arthritis, scoliosis etc. Patients should be counseled that breast reduction will help with relieving pain symptoms caused by excess breast weight. Lastly, hEDS can present with heterogeneous clinical manifestations so careful deliberation and collaboration is needed when deciding on surgery for an adolescent with EDS.

Conclusions

A diagnosis of hEDS should not be considered an absolute contraindication to reduction mammoplasty in patients with macromastia. Because of their predisposition to ailments associated with hypermobility and tissue fragility, patients with hEDS may be more prone to the psychological and physical symptoms of macromastia. Therefore, these patients may benefit more from surgical management. Careful consideration should be given to this patient population when deciding whether to offer them surgery.

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Footnote

Reporting Checklist: The authors have completed the CARE reporting checklist. Available at https://amj.amegroups.com/article/view/10.21037/amj-22-61/rc

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Conflicts of Interest: All authors have completed the ICMJE uniform disclosure form (available at https://amj.amegroups.com/article/view/10.21037/amj-22-61/coif). The authors have no conflicts of interest to declare.

Ethical Statement: The authors are accountable for all aspects of the work in ensuring that questions related to the accuracy or integrity of any part of the work are appropriately investigated and resolved. All procedures performed in this study were in accordance with the ethical standards of the institutional and/or national research committee(s) and with the Helsinki Declaration (as revised in 2013). Written informed consent was obtained from the patient for publication of this case report and accompanying images. A copy of the written consent is available for review by the editorial office of this journal.

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