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A Rare "Big" Problem: A Case Study of Gestational Gigantomastia

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Authors' contributions

This work was carried out in collaboration between both authors. Both authors read and approved the final manuscript.

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Case Report

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ABSTRACT

Aim: Gestational gigantomastia (GG) is an extremely rare condition, with an estimated global incidence rate of approximately 1 in 100,000. This case report aims to examine possible aetiologies of GG and discuss treatment strategies, with a focus on managing its symptoms and achieving a safe pregnancy to full term.

Presentation of Case: We report a case of a 32-year old female at 37 weeks of gestation who presented with rapidly increasing bilateral breast enlargement accompanied with severe back pain, resulting in significant functional impairment. Breast ultrasonography showed heterogenous breast parenchyma with a well-defined heterogenous breast lesion in the right breast. On examination, both breasts were grossly enlarged, tense, tender, with excoriation of the infra-mammary folds. Foetal assessment showed intrauterine growth retardation (IUGR), however amniotic fluid index (AFI) was within normal range. The initial management strategy was for early elective delivery. However, the patient entered spontaneous labour and delivered a healthy baby boy with a normal birth weight. During the immediate post-partum period, there was marked reduction in size of both breasts and significantly improved symptoms.

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Discussion: The aetio-pathology of GG is not yet fully understood and is hypothesized to be linked to hormonal imbalance, auto-immune diseases or malignancy. The management of this condition should be focused on symptomatic relief, prevention of complications in order to carry the pregnancy to term safely and to prevent recurrence in future pregnancies.
Conclusion: Although GG poses minimal risk to the unborn foetus, it can significantly impact the mother's physical and psycho-social well-being. Early referral to a specialised centre is crucial to effectively manage this rare condition, and optimise maternal and foetal outcomes.

Keywords: Gestational gigantomastia; gestational breast hypertrophy; enlarged breast in pregnancy; benign breast disorder; pregnancy.

1. INTRODUCTION

Gigantomastia is a rare condition characterized by diffuse and excessive breast growth (Dancey et al., 2008), while gestational gigantomastia (GG) or gravidic macromastia is a subtype of gigantomastia that arises in pregnancy, typically during the first or second trimester (Mangla and Singla, 2017). It is a disorder that is characterized by diffused, extreme and incapacitating enlargement of one or both breasts during pregnancy (Mangla and Singla, 2017); and is often accompanied with enlarged nipple and areolar size, and prominent superficial veins over the anterior chest (Dancey et al., 2008). It is not known to harm the developing foetus but can have a significant impact on the mother's physical and psycho-social well-being (Alhindi et al., 2023; Qin et al., 2020; Rakislova et al., 2020). We report a case of GG who presented to us at 37 weeks of gestation with bilateral breast enlargement in pregnancy.

2. CASE PRESENTATION

A 32 year old Indonesian female, gravida 3, para 2, at 37 weeks of gestation presented with rapidly increasing bilateral breast enlargement for the past 8 months. She also complained of associated bilateral breast discomfort and severe back pain which had significantly limited her mobility and activities of daily living (ADL). She had initially presented to a primary healthcare clinic at 19 weeks of gestation with bilateral breast enlargement since the third week of pregnancy. Her pre-morbid bra size was a C cup , but as both her breasts rapidly increased in size, she was unable to wear any supportive undergarments. The increase in weight of her excessively large breasts had caused further strain to her back and rendering her mostly bed-bound. She also complained of pain over the infra-mammary folds due to the increase in skin friction and felt social discomfort and self-consciousness to go out in public due to her grossly enlarged breast size. Otherwise, she did not have fever or nipple discharge. There was no loss of weight or loss of appetite; and no family history of malignancy. She is an unemployed foreigner from Indonesia, married to her husband who is of Indian nationality and works earning a minimum-wage. Thus, she is subjected to foreigner hospital fees which is much more costly. This is her first pregnancy of her second union, with no prior history of abnormal breast growth or other complications in her previous two pregnancies. Her last childbirth was more than 8 years ago. She achieved menarche at 14 years old and has a regular menstrual cycle. She had previously used hormonal contraception for four years and practiced turmeric liquid consumption as part of traditional Indonesian practice, known as "jamu". Turmeric-based "jamu" is deeply rooted in Indonesian culture and is believed to promote overall health. They are sold commercially in health stores and supermarkets, and are commonly taken by Indonesian women as part of their daily routine (Rahmat et al., 2021). A provisional diagnosis of Phyllodes breast tumour was made and the patient was given an outpatient appointment to the Breast & Endocrine Clinic. However, she was unable attend her appointment due to financial constraints as she could not afford the clinic consultation fee. She finally presented to us at 37 weeks of gestation due to worsening of symptoms and concerns of her pregnancy.

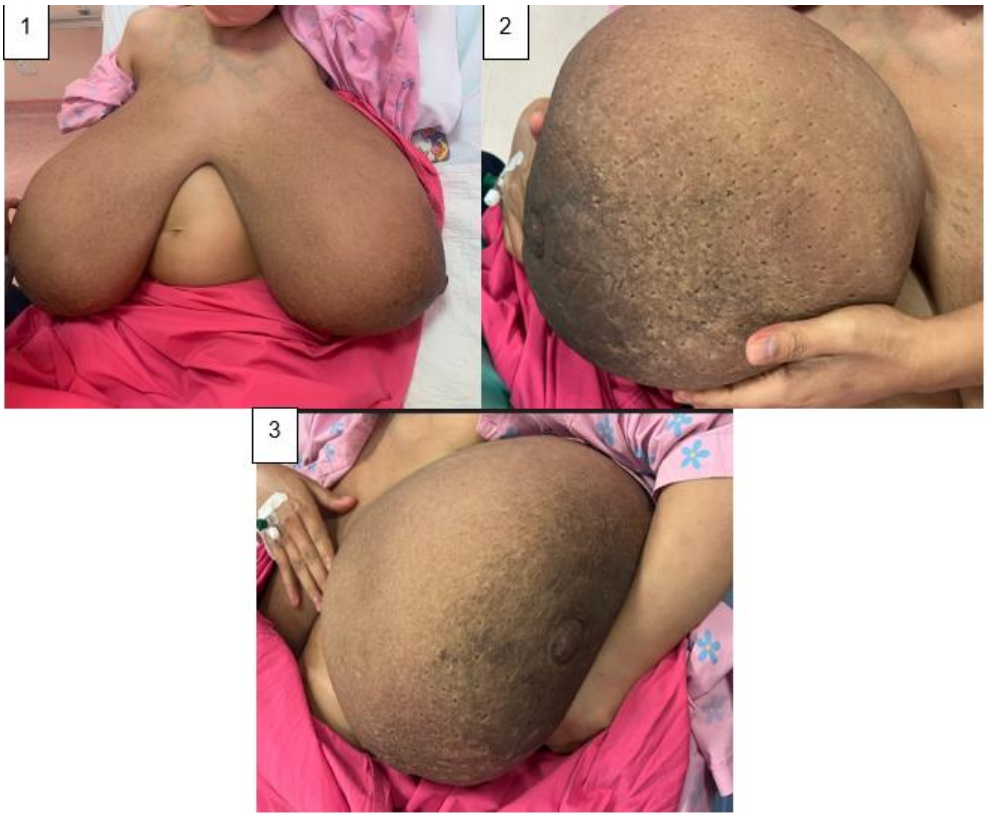
Upon examination, she was alert, clinically pink with good pulse volume and hydration. Vital signs were normotensive, not tachycardic and afebrile. Her bilateral breasts were grossly enlarged, extending beyond the umbilical level. There was peau d'orange skin appearance and dilated superficial veins over the anterior chest (Figs. 1-3). Both breasts were tense and tender on palpation. There were no palpable breast or axillary lumps. Examination of the inferior aspect of bilateral breasts revealed superficial excoriation of the infra-mammary folds, but there

were no skin ulcerations or wounds. No other lymph nodes were palpable.

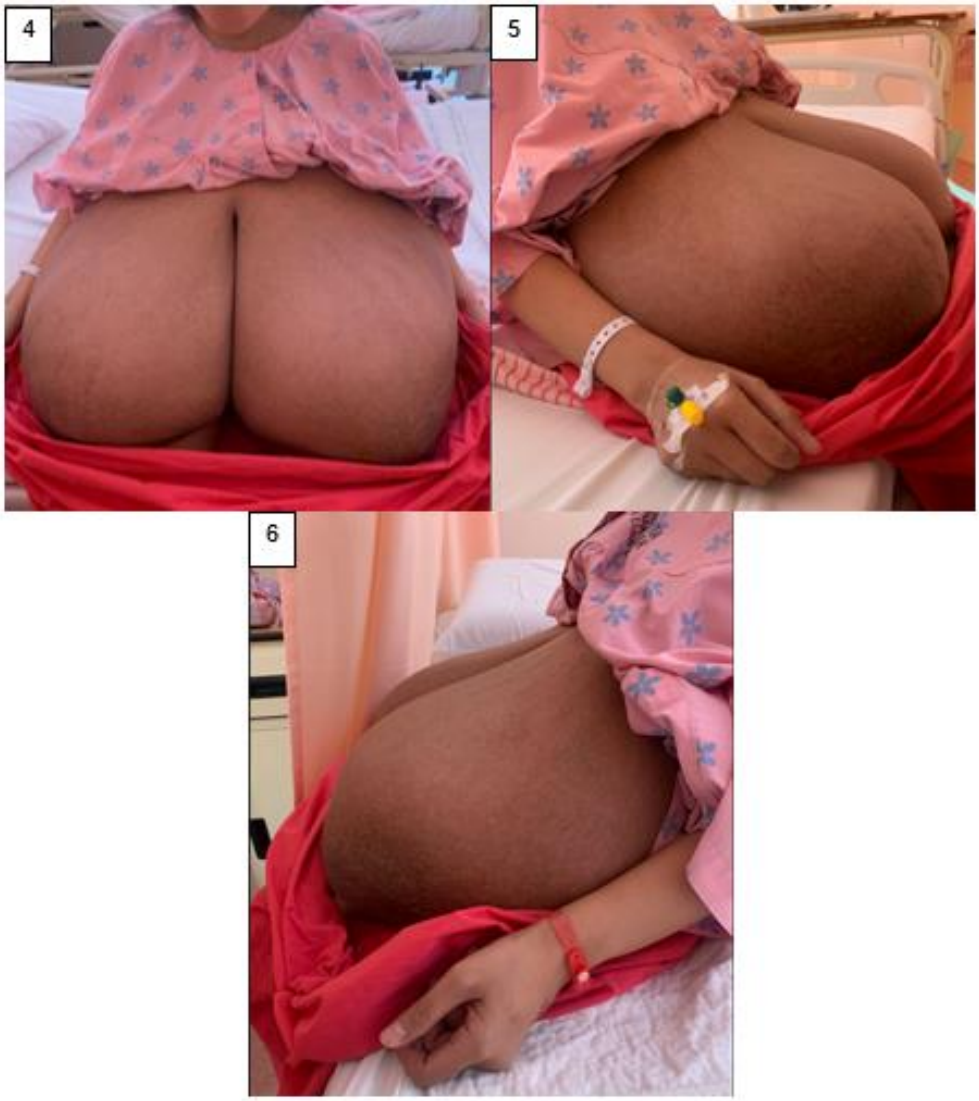
Full blood count, renal profile and electrolytes were all within normal range. Breast ultrasonography demonstrated diffused heterogenous hyper-echogenicity of bilateral breast parenchyma and dilated ducts. There is a well-circumscribed, round, hypoechoic lesion with edge shadowing at the upper inner quadrant of the right breast, measuring 3.3 x 4.4 x 3.4cm (AP x W x CC), BI-RADS 3. There were no enlarged axillary lymph nodes bilaterally. Foetal assessment showed evidence of intrauterine growth retardation (IUGR), but the amniotic fluid index (AFI) and abdominal circumference were within normal range. She was advised for further hormonal testing and fine needle aspiration cytology of the right breast lump, however refused due to the financial concerns on the cost of the laboratory tests.

She was admitted into the Obstetrics ward for pain management and was planned for early delivery via induction of labour. However, she entered spontaneous labour spontaneous at 38 weeks of gestation and delivered a healthy baby boy vaginally, with a birth weight of 2.3kg. At two days post-partum, there was a significant change in the appearance of the bilateral breasts (Figs. 4-6). Both breasts were less tense and had markedly reduced in size. The previously-seen dilated superficial veins over her anterior chest had resolved. She had significantly less breast pain and back pain; and was able to ambulate with ease and carry her newborn baby. Both mother and child were healthy upon discharge to home at three days post-partum.

Medical therapy was not offered as she had presented late in pregnancy. A follow-up review was planned for her in the post-partum period to assess her symptoms and discuss the option of surgery to prevent recurrence. However, she did not attend her appointment again due to financial constraints. Attempts to contact her were futile as the contact number and address given were incorrect.



Figs. 1-3. Bilateral breast swelling with dilated superficial veins at 37 weeks of gestation. (1) Bilateral breasts, anterior view. (2) Right breast, medial view. (3) Left breast, medial view



Figs. 4-6. At post-partum Day 2, bilateral breasts appeared less tense with significant reduction in size and resolved dilated superficial veins over overlying skin. (4) Bilateral breasts, anterior view. (5) Right breast, lateral view. (6) Left breast, lateral view

3. DISCUSSION

Gigantomastia can be classified into 4 subtypes based on aetiology: juvenile, idiopathic, gestational and drug-induced (Fletcher et al., 2020). It is a rare condition with an incidence rate of 1 in 100,000 globally (Dancey et al., 2008). It was first reported by Palmuth in 1684 (Palmuth, 1648) with only 50 published case reports from 1976-2016. Demographic distribution shows that gestational breast hypertrophy is more prevalent in North America, followed by South East and Central Asia (Mangla and Singla, 2017). Cases have been reported in patients aged 16-35 years old, with the majority presenting at 26-30 years old (Mangla and Singla, 2017; Dancey et al., 2008; Türkan et al., 2016; Okere et al., 2023), which corresponds to our patient's age.

The aetio-pathology of this condition is not yet fully understood. However, it has been hypothesized to be associated with systemic disorders such as hormonal imbalance, auto-immune diseases or malignancy (Sanli, 2024). Published case reports have linked GG to hyperprolactinaemia, hypercalcaemia, deranged liver function, myasthenia gravis, anti-phospholipid syndrome and lymphoma (Hodgkin's and non-Hodgkin's lymphoma and T-cell lymphoblastic lymphoma) (Mangla and Singla, 2017; Rezai et al., 2015). Another theory that has been proposed is related to hormone

receptor hypersensitivity or excess in circulating hormones, associated to hormonal changes during pregnancy, such as oestrogen, progesterone, prolactin, testosterone and cortisol (Mangla and Singla, 2017; Fletcher et al., 2020; Türkan et al., 2016). When unrelated to malignancy, GG is a benign condition with no known threat to the foetus in-utero. However, it carries a significant social, emotional and physical disability to the expectant mother. The excess weight and size of the breasts can remarkedly limit their movement, causing significant pain and ulcerations. If poorly treated, infected wounds may lead to sepsis and death (Mangla and Singla, 2017; Okere et al., 2023; Sanli, 2024; Rezai et al., 2015). Our patient was not known to have autoimmune diseases or malignancy, therefore hormonal abnormalities could have been a possible cause. Unfortunately, this could not be proven as she refused for further laboratory testing due to financial constraints.

During the intra-partum period, her foetus showed evidence of IUGR and was born with a birth weight below the 10th centile (WHO, 2006). However, her baby was healthy without any congenital anomalies, and did not require any special neonatal care. In contrast, the patient had suffered great physical and social disability due to the weight of her rapidly-growing breasts that had made her a social recluse and severely limiting her daily activities. Fortunately, she did not have any ulcerations or wounds that could have led to infections, though there were excoriation of the inferior mammary folds as a result of continuous friction which caused her additional pain.

Management of this condition is focused on symptomatic relief, prevention of complications in order to carry the pregnancy to term safely and to prevent recurrence in future pregnancies. The first line of treatment is medical therapy in the early stages with the use of bromocriptine. It is a D2-agonist that arrests further hypertrophy of the breast tissue (Mangla and Singla, 2017). It is safe to use in the gestational period and has not been linked to abortion or congenital anomalies (Krupp and Monka, 1987). However, there have been isolated cases of bromocriptine use that report IUGR, therefore foetal growth should be serially monitored on a regular basis (Rezai et al., 2015). Other options include pain relief with analgesics and the management of any wounds, if present. Elective termination of a viable foetus is not ethical nor indicated as this condition rarely affects the foetus and this does not guarantee a cure.

Another option is surgical management to surgically remove the excess breast tissue. However, this involves a multi-disciplinary approach to plan the surgery as late as possible until the foetus is viable, or to delay until the post-partum period. If urgently indicated, an earlier elective delivery may be planned with the administration of corticosteroid for foetal lung maturity. Most cases do not spontaneously resolve after delivery thus, surgical treatment is offered in the post-partum period as definitive therapy to prevent recurrence in future pregnancies (Mangla and Singla, 2017). Options include reduction mammoplasty and mastectomy with or without breast reconstruction (Mangla and Singla, 2017; Moazzami et al., 2016). Mammoplasty gives the patient the choice for breastfeeding in the post-partum period, which is essential in developing countries. However, it does not eliminate the risk of recurrence as the remaining breast tissue may undergo hyperplasia in subsequent pregnancies. For patients who have a desire for future pregnancies, bilateral mastectomy and breast reconstruction can be considered (Moazzami et al., 2016).

Fortunately, her symptoms showed spontaneous improvement almost immediately after delivery. She did not require medical therapy and was able to deliver safely a healthy baby without major complications throughout her pregnancy. However, the challenge we encountered with our patient is compliancy to seeking medical attention due to her financial difficulties. Her symptoms had appeared at 2 weeks of gestation, but she only presented at 19 weeks of gestation to a primary healthcare clinic, and then defaulted until 37 weeks of gestation to a tertiary hospital. If she had presented earlier, medical therapy could have been initiated to prevent further increase in breast size and help reduce her symptoms. Proper work-up could also have been done to determine the pathological cause and prevent her condition from worsening, or recurring in the future. We are also unable to determine the long-term outcome for her due to lost in follow-up; whether her condition had completely resolve and, or if, it recurs in future pregnancies.

4. CONCLUSION

More awareness must be made of this rare condition amongst primary healthcare doctors

and obstetricians for early referral to a specialised centre, as it requires a multi-disciplinary management strategy. Further research is needed to better understand and treat this condition to attain optimal foeto-maternal outcome.

DISCLAIMER (ARTIFICIAL INTELLIGENCE)

Author(s) hereby declare that NO generative AI technologies such as Large Language Models (ChatGPT, COPILOT, etc) and text-to-image generators have been used during writing or editing of this manuscript.

CONSENT

Both authors declare that informed consent was obtained from the patient for publication of this case report and accompanying images.

ETHICAL APPROVAL

Both authors hereby declare that all measures taken for the completion of this case report are in accordance to the 1964 Declaration of Helsinki.

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COMPETING INTERESTS

Authors have declared that no competing interests exist.

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