



Breast Developmental Anomalies: A Review of the Problem

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Author's contribution

This work is a sole-authorship one. The author designed the review, wrote the protocol and wrote the draft of the manuscript and managed the literature searches; diligently read and approved the final manuscript.

Review Article

Received 9th June 2012
Accepted 23rd August 2012
Published 16th October 2012

ABSTRACT

The breast is a sign of female sexual identity. However, there are some anomalies that affect normal breast development. These abnormalities could have physiological, psychological and psychosocial effects on an individual and could result in the person developing a negative self image. Not much study on these anomalies has been carried out even though most people are likely to suffer from any one of these breast developmental abnormalities (BDA) than breast cancer. Awareness on these abnormalities is very low unlike breast cancer. Due to low awareness, people with this form of condition are not aware of measures available. Incidence of these anomalies could be high hence the need to educate people on these abnormalities. This review seeks to create the awareness on BDAs and the necessary interventions available.

Keywords: *Breast developmental anomalies; breast cancer; awareness; physiological effect, psychological effect; psychosocial effect.*

ABBREVIATIONS

Breast Developmental Anomalies (BDA); Reduction Mammoplasty (RM).

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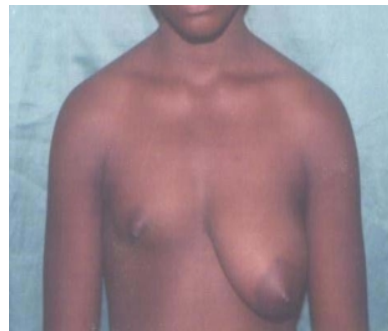
1. INTRODUCTION

The female breast refers to the mammary gland which provides milk to nourish a baby in women and also a sign of female sexual identity. It is the origin of the most common malignancy (breast cancer) in women (Siegel et al., 2011). Embryologic development of the mammary gland consists of a series of highly ordered events involving interactions among a number of distinct cell types. These interactions are regulated by an array of systemic and local factors such as growth factors and hormones. At 6 weeks, mammary glands begin to develop in the embryo when an ectodermal ridge begins as a bilateral thickening extending from the axilla to the groin. Although most of this eventually atrophies, small bilateral portions in the pectoral region remain and these become the breast (Seltzer, 1994). The breasts are situated superficial to the pectoralis major muscle and are hemispheric in shape with an elliptical base in the average young woman extending between the second and sixth ribs, vertically and horizontally between the lateral edge of the sternum and midaxillary line (Shermak, 2010). The breast consists of glandular tissue, fibrous tissue and fatty tissue with each breast consisting of 15-20 lobes of milk secreting glands embedded in the fatty tissue.

Breast development occurs usually between the ages of 8 and 13 as a result of increased oestrogen levels. The mammary gland in males does not undergo post natal development unlike females in which development of breast (thelarche) is a secondary sexual characteristic (Neinstein, 1999). Due to menstruation, pregnancy, lactation and menopause, various changes occur in females resulting in variation in breast size, shape and appearance. Hormones which influence breast development include: Transforming growth factor alpha (TGF- α) which stimulates ductal and lobulo-alveolar development; TGF- β which affects canalization of ductal structures and suppression of lactation (Imagawa et al., 2002). IGF-1 impacts ductal growth and is expressed in mammary stroma (Imagawa et al., 2002; Loladze et al., 2006). Other hormones include prolactin (a hormone produced by the pituitary gland which helps to stimulate the development of the mammary gland and initiates and maintains milk production), oestrogen (responsible for female sexual development; excess may cause gynaecomastia in men) and progesterone (responsible for the functioning of the female reproductive system). Both progesterone and oestrogen cause milk producing glands to develop during pregnancy. Breast developmental anomalies could either be classified as over-development or under-development of the breast. Under-development includes: Hypomastia/hypoplasia (Fig. 1), aplasia/amastia (absence of breasts). Over-development includes: Polymastia (extra breasts), macromastia, gigantomastia, polythelia.



(a) Bilateral Hypoplasia in a 16 year-old girl



(b) Unilateral Hypoplasia in a 17 year-old girl

Fig. 1. Hypoplasia (Source: Author)

Another form of BDA is Tubular/tuberous breast (breast shaped like a tube) (Fig. 2).



Fig. 2. Tuberous/Tubular breasts in a 15 year-old (Source: Author)

There are also forms such as Polythelia (extra nipple or nipple-areolar complexes) (Fig. 3) and also malformations of the nipple (Fig. 4).



Fig. 3. Bilateral accessory nipples (Source: Author)



Fig. 4. Nipple malformation: Split-nipples (Source: Author)

Athelia (absence of nipple), Poland Syndrome (a rare birth defect characterized by underdeveloped or absent chest muscle, associated with hypomastia), Breast asymmetry (unequally sized breasts) are some other forms of BDAs. Another form of over-development BDA is Juvenile/Gestational Gigantomastia/Macromastia (massive hypertrophy of the breast caused by either increased sensitivity to the female hormones, oestrogen and progesterone or elevated levels of the hormones in the blood) (Fig. 5A & 5B).

Gynaecomastia is a condition which occurs in males as a result of oestrogen excess causing their breasts to appear as females.

Some of the BDAs may have unknown aetiology (Howard and Gusterson, 2000), while others are caused by hormonal imbalance. Amastia and athelia result when the mammary ridges fail to develop or completely disappear (Arca and Caniano, 2004). Polythelia occurs in 1%-2% of the population (Greydanus, 2006), often unilateral with 95% of the supernumerary nipples occurring along the milk line (Coffin, 2002). Polymastia and Polythelia occur much more in females than males (Loukas, 2007). Tuberous breasts could be congenital

(Panchapakesan and Brown, 2009) and could range from mild to severe. Breast asymmetry occurs when there is sagging or drooping, a natural phenomenon at menopause. However, sagging could be congenital in some people when the suspensory ligaments loosen up and is not able to support the breast.



Fig. 5A. Juvenile macromastia (Source: Author) **Fig. 5B. Macromastia (Source: Author)**

Poland Syndrome is the most frequent cause of breast hypoplasia or aplasia; the right side is often affected than the left with men more frequently affected than women to the ratio of 3:1 (Borschel et al. and da Silva Freitas, 2007). Gynaecomastia is the commonest form of breast hyperplasia occurring in 30-57% of healthy men (Lanitis et al., 2008). Gynaecomastia could occur as a result of delay on testosterone secretion leading to greater oestrogen effect. Gestational gigantomastia is a rare connective tissue disorder estimated to occur in 1 out of 28,000 to 100,000 pregnancies worldwide (Dancey et al., 2008).

2. PREVALENCE OF BREAST DEVELOPMENTAL ANOMALIES IN PARTS OF GHANA

Even though BDA is a necessary worry, not much study has been done. Agbenorku et al. (2010) conducted a study at Sogakope, Ghana which revealed that 20.7% of 550 female students suffered from one of the BDAs. A retrospective study conducted by Agbenorku et al. (2007) in Kumasi, Ghana also showed 38 cases of BDA with macromastia (47.37%) being the commonest form of the condition reported. This shows that prevalence could be high in some parts of the country, hence the need for further studies to be carried out.

3. EFFECTS OF BREAST DEVELOPMENTAL ANOMALIES

3.1 Physiological

Extremely large breasts cause not just emotional distress but also physical distress such as pain to the neck, back, shoulder, head and in some cases poor posture causing the upper spine to curve forward, a condition known as kyphosis. According to the Plastic and Craniofacial Surgery for Infants and Children (Chrominski et al., 2003), excessively large breasts can even cause hand and finger numbness.

Some individuals with breast hypertrophy (extremely large breasts) develop grooves on their shoulders from the weight of their bra straps and some difficulty in wearing some kind of clothes. Pathological changes in the vertebral column as a result of gigantomastia cause discopathia, scoliosis or scoliokyphosis (Chrominski et al., 2003).

Tuberous breast also poses some discomfort to individuals such that in some cases the individuals cannot breast feed while in others, the milk glands are not developed to produce milk.

3.2 Psychosocial

Breast development occurs usually between the ages of 8 and 13 as a result of increased oestrogen levels and is usually one of the first signs of puberty. However abnormal breasts may cause worry to the teen and sometimes the family of the affected person. It could also result in embarrassment preventing the individual from engaging in some activities of which the person was one active. The female breast is an important component of her body's image. BDA can cause an individual to develop low self esteem since it is a sign of femininity and sexuality. It could also result in isolation and may cause the individual sometimes to feel embarrassed among his or her peers and therefore would not want to associate with them. If awareness is made known and necessary intervention put in place, this ill feeling could be avoided. Tadaoki et al. (1993) reported a case of a 12-year old girl with juvenile hypertrophy of breast which caused her intense psychological problems, incapacitating her in school activities and social relations.

4. AWARENESS OF BREAST DEVELOPMENTAL ANOMALIES

Due to inadequate information on BDA most girls and even boys who have this condition are unaware about some treatment measures available. The level of awareness of this anomaly nationally is low unlike breast cancer, one of the leading causes of death in women across the globe.

Only few studies have been conducted in the country, Ghana. Agbenorku et al. (2011) conducted a similar study to determine the level of awareness of BDAs in Jamasi, Ashanti Region of Ghana. Out of 600 female students, 83% were aware of the condition meaning the awareness in this community was high unlike Sogakope in the Volta Region where awareness was on the low side. Agbenorku et al. (2010) concluded that 80% of the participants in Sogakope said they would accept to go for treatment when made readily available. One could conclude from the study that BDA incidence could be high in several communities; hence the need for awareness programme put in place as is done about breast cancer in order to educate the general public.

5. DIAGNOSIS

Cautiousness is needed in diagnosing breast disease and breast anomalies so that wrong diagnosing is avoided and appropriate care and attention given to individuals presenting with either a malignant or benign breast disease.

Somma et al. (2012) stated that, due to the potential malignant transformation of the abnormal breast, early and accurate diagnosis is required as well as its monitoring over time. Some benign breast diseases such as atypical hyperplasia are risk factors to the individual

developing breast cancer at a later stage in life; therefore breast examination needs to be carried out on patients during antenatal visits. According to Ader et al. (1997), any complain of breast pain must be handled very well in order to determine if it is as a result of a physiological or pathological process.

Gynaecomastia is a benign proliferation of the glandular tissue of the male breast; it may or may not be associated with pain (Shermak, 2010). It could be unilateral or bilateral, examined as a palpable mass of tissue at least 0.5cm in diameter underlying the nipple, hence must be carefully differentiated from breast carcinoma (Braunstein, 2007).

5.1 Use of Ultrasound

Ultrasound imaging is ideal in evaluating breast lesions to prevent a later deformity caused by surgery. Ultrasound can help evaluate breast masses in children to prevent misdiagnosis as most breast lesions in children and adolescents are usually benign. It is the ideal imaging modality to study the pediatric breast and can be useful in all cases in identifying and characterizing the abnormality (García et al., 2000). Ultrasound for diagnosis of gynaecomastia is recommended since asymmetric gynaecomastia in boys may be mimicked by general obesity and pectoral hypertrophy.

6. TREATMENT

6.1 Surgical Procedures

Different treatment options are available depending on the kind of anomaly the individual presents. Proper timing of surgery is necessary to produce good results. Great caution must be exercised when creating incisions around the prepubertal breast. In children, breast malignancies are rare hence there must be critical observation and analysis before a biopsy of a suspicious mass is taken (Shermak, 2010).

In gynaecomastia, surgery may be carried out to remove excess breast tissue. Laituri et al. (2010) in a study on treatment of adolescent gynaecomastia concluded that milder forms can be managed by subcutaneous mastectomy and the severe forms by reduction mammoplasty. Gynaecomastia is usually treated with liposuction or liposuction with excision of the glandular tissue.

Zagara et al. (1999) reported a case of a 30 year old woman who presented with unilateral gestational macromastia which required reduction mammoplasty.

Tuberous breast can be treated by surgical procedures using tissue expansion methods and/or combined with mastopexy (Versaci and Rozzelle, 1991).

Surgery could also be carried out in polythelia. Some anomalies may require breast augmentation with implant, mastopexy (breast lift), and nipple size adjustment. Chrominski et al. (2003) also recommended reduction mammoplasty for kyphosis and also that surgery is delayed until the end of puberty when breast growth is complete. Surgery produced excellent result in a 51-year old female patient who presented with gigantomastia (Chrominski et al., 2003). Antevski et al. (2011) reported a case on extreme gigantomastia in pregnancy of a 30 year-old woman with a tissue of 20kg removed by reduction mammoplasty.

6.1.1 Reduction Mammoplasty (RM)

RM or Breast Reduction involves the reduction of the size and volume of breast by the removal of excess fat and skin. The remaining breast tissue is tightened and repositioned to create breast that best suits the patient's body. Most surgeons recommend that the procedure be carried out after the breast is fully developed and some also recommend that surgery is carried out after the woman is done with childbirth since pregnancy and breast feeding could have some effects on the size and shape of breasts. Incision is made which could be a micro-incision (if the enlargement is mainly due to fats and not the skin), peri-areola incision (if the incision is around the perimeter of the areola), lollipop incision (vertical scar technique) and anchor incision (inferior pedicle flap); the surgeon chooses the type of incision that best suits the patient. This is followed by the removal of the excess breast tissue and the remaining tissue reshaped. The nipple and areola are also re-positioned. The incision is then closed and the remaining skin tightened. Sutures are layered deep within the breast tissue to support the newly shaped breasts. Surgical tape and skin adhesives could also be used to close and support the skin (Zargara et al., 1999).

6.1.2 Mastopexy

Mastopexy helps to reduce enlarged areola sizes, to elevate the nipple to a more anatomical position and to restore shape and firmness. This procedure is usually carried out on women who have ptosis from significant weight loss or postpartum ptosis. Mastopexy cannot significantly reduce the size of the breast and hence performed alongside breast augmentation or reduction. Incision is made depending on the amount of excess skin and the position of the nipples. The breast is repositioned and reshaped and the excess tissue excised. The nipple and areola are also re-positioned. The incision is then closed and the remaining skin tightened. Sutures are layered deep within the breast tissue to support the newly shaped breasts. Surgical tape and skin adhesives could also be used to close and support the skin (Lanitis et al., 2008).

6.1.3 Breast Augmentation

This involves the use of implants to restore breast volume lost as a result of massive weight lost or pregnancy affecting the size and shape of the breast. This is usually carried out on persons who feel their breasts are out of proportion to the rest of their body. The incision is made at an inconspicuous area; these incision options could be inframammary, periareolar, transumbilical or transaxillary. Choice of incision type is based on the type of implant (silicone or saline breast implant; depends on breast anatomy, body type or skin elasticity), degree of enlargement, anatomy of patient and the surgeon's preference. Breast implants can be placed in one of the two locations: subglandular (on top of the pectoral muscle) or subpectoral (below the pectoral muscle). Incisions are closed with layered sutures in the breast tissue. Surgical tape and skin adhesives could also be used to close the skin (Lanitis et al., 2008).

6.2 Psychological Management

Negative self image resulting from breast development anomalies is very common. Counseling is needed to help the individual to do away with any ill-feeling such as depression resulting from their body image. Therapy helps these individuals to have a positive self image; peers and family members can help these individuals to be able to be active once again in activities they enjoy. Also, health professionals could also educate

persons on interventional measures available to help persons presenting with any of these forms of the abnormality. Kinsella et al. (2012) recommended psychological evaluation and treatment as an adjunct to successful surgical management of gynaecomastia.

6.3 Medical Management

Even though, medication has not been really effective in the management of breast development anomalies, they could be used as adjunct to surgical procedures as occurred in 12- year girl suffering from juvenile hypertrophy who was given tamoxifen after the removal 2 kilograms (4.4 lb) of tissue from her right breast and 1.9 kilograms (4.2 lb) from her left breast to suppress breast re-growth (Tadaoki et al., 1993). Chrominski et al. (2003) stated medication such as bromocriptine, progesterone, testosterone and tamoxifen for the treatment of gigantomastia have not been very effective and therefore recommend surgery as a suitable choice.

7. CONCLUSION

The need for education on breast developmental anomalies is necessary. As is done on breast cancer awareness, seminars, symposia and campaigns are some of the means by which the populace could be informed on these conditions. Also love, care and support shown to individuals with any of the anomalies is a necessary tool to help them gain full control of their lives again and gain their self esteem; this would help them to readily seek for treatment.

CONSENT

The author declares that written informed consent was obtained from the patients used in this article.

ETHICAL APPROVAL

The author hereby declares that the appropriate ethical approval was obtained from the Kwame Nkrumah University of Science and Technology School of Medical Sciences/Komfo Anokye Teaching Hospital Ethical Committee and all procedures have therefore been performed in accordance with the ethical standards laid down in the 1964 Declaration of Helsinki.

COMPETING INTERESTS

Author has declared that no competing interests exist.

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