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Amazia: A rare anomaly in a resource poor setting.

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Abstract

Amazia is a condition where one or both of the mammary glands is/are absent in the presence of the nipple and areola. We report the case of a 13 year old girl who presented in our Paediatrics endocrinology clinic with failure of the right breast to develop while there were progressive changes of normal puberty in the left breast and other parts of the body. There were no dysmorphic features reminiscent of any syndrome. Management was hampered by financial constraint.

Keywords: Amazia, Puberty, Resource poor.

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Introduction

Amazia is a condition where one or both of the mammary glands is/are absent in the presence of the nipple and areola [1].This is different from athelia a condition in with one or both nipples are absent and amastia which is a condition where breast tissue, nipple and areolar are absent [1,2].

Amazia can be congenital, acquired or iatrogenic (following surgical removal and or radiation therapy). It can also be bilateral or unilateral. Its incidence is not known possibly because it is a rare disorder which occurs majorly in females and most people don't feel comfortable discussing it or presenting at the hospital because of adverse psychosocial effect of the disorder.

It can be diagnosed during early puberty when there is failure of one or both breasts to develop in the presence of other secondary sexual characteristics.

Amazia can be associated with certain syndrome like Poland syndrome, Mobius Syndrome and klippel-feil syndrome [1]. However, iatrogenic causes of amazia is more common than other causes and this may follow incision and biopsy of breast bud earlier in life, trauma and radiation to the chest wall [1,2].

Case Report

A 13 year old girl presented in our Paediatrics endocrinology clinic with underdeveloped right breast. This was noticed about 1 year before presentation when the left breast started developing and the right breast was noticed not to show any sign of growth. The left breast grew steadily to Tanner stage 3 and the pubic hair was at least stage 2 at the time of presentation. The attention of the aunt was drawn to it by the patient's elder sister who, on visitation, noticed the lack of development of her sister's right breast few weeks before presentation when the patient was undressing in her presence.

Patient has not attained menarche. There has been no similar occurrence in the family. There was no history of trauma or surgery to the right hemi thorax. There was no history of use of oestrogen containing cream on left hemithorax or history suggestive of sexual abuse in the patient. There was no chest wall, hand or palm deformities, and no craniofacial abnormalities. The patient was not ill looking and she had normal weight and height for age and gender. The left mammary gland was well developed with breast tissue, areola and normal nipple. The right hemithorax had a well-developed nipple and areola in the normal anatomic position for breast but no visible or palpable underlying breast tissue (Figure 1). Laboratory investigation and possible plastic surgery was hampered

Figure 1. *Patient with Amazia*

by gross financial constraint. Patient's anxiety was allayed and she was counselled to pad her brassier on the right to avoid unnecessary embarrassment from school mates until funds will be available for definitive management.

Discussion

Breast development starts as early as 4 to 6 weeks of gestation when mammary specific progenitor cells may be seen [3]. Around day 35 of gestation, proliferation of paired areas of epithelial cells in the epidermis of the thoracic region occurs. These discrete areas of proliferation extend in a line between the foetal axilla and inguinal region and form two ridges called the mammary crests or milk lines. Most of the mammary milk line atrophies except for paired solid epithelial masses in the pectorial region at the fourth intercostals space, which form the primary mammary bud. Supernumerary nipples occur in 2 to 5% of human along the milk line [4,5]. At the end of first trimester a well defined mammary bud penetrating into the upper dermis can be observed [6]. By the second trimester, secondary epithelial buds appear from the indentations on the main mammary bud [4,6,7]. By the 6^{th} month of gestational age, the basic framework of mammary gland is established and breast tissue can be apparent in male and female foetuses at this stage [8]. Repeated branching of the secondary epithelial buds and canalization occur by the third trimester [6,7]. During the final weeks of gestation, there is increased vascularisation of loose fibro-connective tissue of the mammary gland and limited milk secretion may occur in the breast of term newborn due to complex interplay between foetal, placental and maternal hormones [7,9]. Absence of breast nodules and milk secretion in the breasts of preterm babies indicate that the intrauterine environment is essential for breast development [10,11]. Amazia is thought to be secondary to a disruptive sequence involving the braciocephalic arterial system in early embryonic life [1].

Amazia is a known component of certain syndromes. Poland Syndrome involves breast anomalies with a range from total amastia to athelia, varying degrees of underlying chest wall depressions and unilateral brachysyndactyly [12,13]. Familiar cases of Poland syndrome has also been reported [14]. Trier et al. [15] noted that bilateral absence of the breast was associated with some craniofacial anomalies. Amazia has also been associated with congenital bilateral choanal atresia [16]. Golden-ring, Crelin et al. [17] reported a mother and daughter with bilateral absence of the breast, sparse axillary and pubic hair, saddle nose, hypertelorism and high arched palate. This also suggests a hereditary pattern.

In the index patient there were no craniofacial abnormalities or limb abnormalities and no family history which could suggest a possible hereditary pattern. Also, there was no history of trauma, surgery or radiation to the right hemithorax previously.

Management of amazia is usually by surgical reconstruction of the breast with breast implant [2,18]. Most patients cannot afford this treatment due to its high cost and there may also be non-availability of necessary facilities especially in developing countries like ours and therefore patients are prone to depression or loss of self-esteem as teenagers and may later have problems feeding their young ones in a society where childhood survival is majorly made possible through breastfeeding especially in bilateral amazia. Alternative to breastfeeding in such cases will include breast milk substitute, wet-nursing or breast milk bank, wherever such is available and culturally acceptable. Patients with unilateral amazia, apart from cosmetics and psychological challenges, should usually be able to breastfeed their babies on one breast with good social support.

More research will be of great important to determine the aetiology of amazia with the view to preventing its occurrence. There is also need for collaboration and surgical assistance from highly specialized medical institutions to assist patients with this and similar conditions.

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