

# Developmental anomalies of the breast in the pediatric population

 Mustafa Yaşar Özdamar

Department of Pediatric Surgery, Faculty of Medicine, Erzincan Binali Yildirim University, Erzincan, Türkiye

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Corresponding Author: Mustafa Yaşar Özdamar, mustafayasarozdamar@gmail.com

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## ABSTRACT

Pediatric breast conditions, which rarely require biopsy or excision and where follow-up with ultrasound is sufficient, are usually temporary. To understand childhood's physiological and pathological conditions, it is necessary to know the stages of intrauterine development, as in every organ. In neonatal and subsequent childhood periods, ultrasound is sufficient as a radiological examination after history and physical examination. Malign masses are sporadic, and benign masses are rarely encountered. Parental anxiety is an essential factor affecting patient management from the first admission to the final follow-up examination. For this reason, practitioners need to know information that reassures patients and parents with an appropriate methodology in developmental breast diseases.

**Keywords:** Children, breast, developmental anomalies

## INTRODUCTION

From the fifth week in the embryological period, the milk streak (mammary ridge) develops bilaterally in the ectodermal layer along the trunk from the axilla to the groin. Although many paired mammary glands develop along this ridge, they disappear shortly after formation, except for a portion in the anterior fourth intercostal space. Until birth, solid epithelial columns comprising 15-20 branches with specialized epithelium canalize to form lactiferous ducts. The ducts continue to form lobules, which continue until the formation of a solid dermal button that will form the nipple between 7 and 12 weeks intrauterine week. Mesenchyme, which will become the connective and fatty tissue supporting the breast, surrounds the epithelial system. Although there is a temporary pit during the epithelial invagination of the nipple from the skin, close to birth, this pit extends outwards and forms the nipple. If there is a disruption at this stage, inverted nipples will rarely occur at birth. The breast consists of lactiferous ducts without alveoli from birth to puberty. Breast development has the same developmental stages in both genders until puberty. During the pubertal period under hormonal influences, the ducts that will form the breast lobules in the future begin to proliferate, and their terminations form solid masses of cells.<sup>1-3</sup>

The thelarche period, when breast development begins in girls at the age of 9-10, represents the beginning of the puberty period. In thelarche, which starts with the completion of the terminal duct lobular unit in the breast, estrogen triggers ductal growth, and progesterone triggers lobular and alveolar differentiation.<sup>4,5</sup>

This stage of breast development concerns the pubertal period, which is the beginning of the first step into adulthood. There are no breast problems that would put the clinician in a problematic situation. For example, thelarche that begins at 7-8 is called the premature thelarche and is seen in prepubertal girls. Thelarche, the beginning of secondary breast development, is considered delayed if it begins after twelve years old. In such a case, if there is no source of hormonal pathology originating from the ovary or adrenal gland, the patient can be followed up by an endocrinologist after this stage.

In the pediatric population, except for developmental breast changes from the neonatal period to adolescence, benign masses occur at rates as low as 3%, especially after puberty. Malignant breast diseases are encountered sporadically at a rate of 1/100000. This study will discuss developmental childhood breast conditions other than rare benign and malignant diseases.

## 1. DEVELOPMENTAL ANOMALIES OF THE BREAST

### 1.1. Polythelia

In the fetus, after the 12th intrauterine week, a residue in the mammary ridge from the axilla to the groin, other than the usual location, causes polythelia (supernumerary nipples). The most common location is just below the breast or on the skin of the abdomen. Incidence is 0.2 to 2.5 percent. This abnormality, seen bilaterally in fifty percent, has been reported to be rarely seen, even in the scapula, head, neck, and thighs. While there is no diagnostic or symptomatic problem with this anomaly, clinicians should remember that it may accompany urological malformation or malignancy.<sup>6-8</sup>

### 1.2. Ectopic Breast Tissue

Ectopic breast tissue, in embryological life, occurs from the residue outside the original breast location, where the mammary ridge is located, as in polythelia. The presence of ectopic or accessory breast is called polymastia. Ectopic breast tissue may contain components of the mammary gland, including the nipple, areola, and glandular tissue, and may be exposed to benign and malignant breast diseases. Ectopic breast, which has a prevalence of 2-6 percent in the literature, constituted 3.2% of our breast patients (17 out of 409) in the last eight years. If ectopic breast tissue is usually in the axillary region and cannot be diagnosed with ultrasound, fine needle aspiration biopsy can be performed. Total excision should be performed if these patients have aesthetic concerns and suspicion of benign or malignant disease.<sup>7,9</sup>

### 1.3. Premature Thelarche

It is an annoying condition in girls under 7.5 years of age, where there is only premature breast growth in the absence of secondary sex characteristics (such as pubic hair, menarche, and axillary hair growth). Regular follow-up is sufficient unless the patient has pelvic ultrasound and biochemical hormone findings indicating precocious puberty.<sup>4,10</sup>

### 1.4. Neonatal Breast Hypertrophy

Although it is accepted in the literature that maternal estrogen is the etiological factor of breast hypertrophy seen in newborns, it has been understood that placental-derived estrogen causes this condition. This is because there is no direct hormone transfer from mother to fetus in intrauterine life. Breast hypertrophy, which is generally seen in seventy percent of newborns, is more common in females, and galactorrhea may be observed in 5% of patients. This situation (galactorrhea) is popularly known as witch's milk. Although ultrasound is performed on such babies due to the family's concern, the finding on the ultrasound indicates that there are only secretory ducts and no lobules, so no negative situation is encountered. In short, hypertrophy, which becomes evident in the first week of life, is temporary.<sup>2,5,9,11</sup>

### 1.5. Hemangioma and Lymphangioma

Pre-adolescent breast hypertrophy may be accompanied by hemangioma and lymphangioma. Even if it is recognized by clinical appearance, the diagnosis can be confirmed with ultrasound and magnetic resonance. Hemangioma and lymphangioma treatment protocols vary from case to case.<sup>1</sup>

### 1.6. Mammary Duct Ectasia in Infantile

This entity, whose etiology is unknown, has been tried to be demonstrated by the presence of large dilated mammary ducts in postmortem studies in healthy one-three-year-old children. Bloody discharge from the nipple is the most common symptom and palpable mass may accompany it. While this disease shows symptoms at this age with mini-puberty symptoms, it may be seconder to inflammation, bleeding, and duct obstruction in the breast in older children. It is more common in the male child population. This disease is temporary, requiring appropriate conservative treatment, and a process that follows with parental reassurance usually suffices.<sup>1,2,12</sup>

### 1.7. Nipple Inversion in Newborn

Its overall prevalence is approximately two percent. This anomaly is caused by the breast ducts remaining shorter than usual during the fetal period and being inverted into the areola with fibrous bands. When such baby girls reach adulthood, they do not experience breastfeeding difficulties due to changes in the breast during pregnancy. Still, this anomaly may need to be corrected with a mini operation.<sup>1,3,13</sup>

### 1.8. Lipomastia

It occurs in obese children due to the increase in fat tissue in the breast. If it cannot be differentiated from gynecomastia during physical examination, a differential diagnosis can be made with ultrasound. The absence of glandular tissue under the areola when the patient is in a sitting position is a helpful factor in diagnosis.<sup>4</sup>

### 1.9. Gynecomastia

Among the robust assumptions that familial factors are influential, Gynecomastia is seen in 4-64% between the ages of 10-13, typically six months after the emergence of secondary sex characteristics. Although this condition disappears around the age of 17, it is a benign process and regresses on average in two years. The etiology of this pathology, which is especially seen in prepubertal boys, cannot always be determined. Exposure to endogenous or exogenous estrogen is the most considered etiological factor. The main ones that should be investigated as endogenous sources are gonadal or adrenal tumors, gonadotropin-secreting tumors such as hepatocellular carcinoma or choriocarcinoma, aromatization of androgens associated with Sertoli cell and sex cord testicular tumors, and Klinefelter syndrome. Estrogenic and antiandrogenic effects of cosmetic products as exogenous sources may carry out gynecomastia.<sup>14</sup> Recently, it has been stated that Leptin, which is found in breast epithelial cells and causes an increase in estrogen concentration by increasing aromatase enzyme activity in adipose tissue and breast tissue, may be effective in the etiology of gynecomastia.<sup>15</sup> Treatment is often not required unless gynecomastia is persistent and severe.<sup>14,15</sup>

### 1.10. Asymmetrical Breast Development and Related Diseases During Adolescence

Although breast asymmetry develops from infancy to adolescence, emotional pathology may occur in adolescents as self-esteem becomes dominant in adolescence. In this case, although gynecomastia in male adolescents is diagnosed through clinical examination, ultrasound can be performed for the reassurance of the family and the patient. If the clinician has suspicious examination findings in marked asymmetry,

extreme diseases such as fibroadenoma should be considered with ultrasound and other examinations. Apart from breast asymmetry and physiological asymmetry in girls with scoliosis, true breast asymmetry may be congenital in cases of amastia, hypomastia, and Poland syndrome.<sup>16-18</sup>

Amastia, which is usually accompanied by other embryological ectodermal defects and where the ridge of the breast does not develop or is completely involuted, also accompanies Poland syndrome. Amastia or hypomastia may be seen in Poland syndrome, which is characterized by the complete absence of unilateral chest wall muscles and ipsilateral rib cage and upper limb defects. Surgical procedures to correct breast asymmetry in adolescents should be postponed until breast development is completed.<sup>16-19</sup>

### 1.11. Adolescent Breast Hypertrophy

This entity, also known as juvenile or virginal breast hypertrophy, refers to sudden and progressive breast growth over a few months. Since it contains histological findings of gynecomastia, it is also called gynecomastoid breast hypertrophy. Antiestrogenic drugs such as tamoxifen are used to stop breast growth in this hypertrophy, which can also be familial and cause embarrassment and back pain in girls. Even if breast growth is partially halted, this situation needs to be corrected with surgery.<sup>17,20</sup>

### 1.12. Fibrocystic Changes in The Breast

This condition, which was known as “fibrocystic disease” until the nineties, is the manifestation of macroscopic and microscopic cysts, apocrine metaplasia, fibrosis, and duct adenosis in the breast, resulting from the imbalance between estrogen and progesterone, and accompanying epithelial hyperplasia in its histopathology.

Especially during pre-menstrual periods, breast tenderness and orange peel-like nodularity are palpable on examination. There are no specific findings on ultrasound other than cysts and fibrosis areas. The patient is followed at regular intervals.<sup>17,21</sup>

## CONCLUSION

In the pediatric population, healthcare providers to whom breast patients consult from the neonatal period to the age of 18 should be knowledgeable about developmental physiological and pathological breast conditions. Because parents experience intense anxiety, thinking that their child may develop breast cancer similar to that seen in adults.<sup>22</sup> Pediatric breast conditions, which rarely require biopsy or excision and where follow-up with ultrasound is sufficient, are usually temporary.

## ETHICAL DECLARATIONS

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## REFERENCES

- Coffin CM. The breast. In: Stocker JT, Dehner LP, eds. *Pediatric pathology*. 2<sup>nd</sup> ed. Lippincott Williams & Wilkins: 2002:993-1015.
- Felsch KN, Merritt DF. Breast concerns. In: Kliegmen RM, Stanton BF, Schor NF, St Geme III JW, eds. *Nelson Textbook of Pediatrics*. 20<sup>th</sup> ed. Elsevier: 2016:2614-2618.
- Harold Ellis, Vishy Mahadevan. Anatomy and physiology of the breast. *Surgery (Oxford)*. 2013;31(1):11-14.
- Kaplowitz PB. Reexamination of the age limit for defining when puberty is precocious in girls in the United States: implications for evaluation and treatment. *Pediatrics*. 1999;104(4):936-941.
- Bock K, Duda VF, Hadji P, et al. Pathologic breast conditions in childhood and adolescence. *J Ultrasound Med*. 2005;24(10):1347-1354.
- Mehes K. Association of supernumerary nipples with other anomalies. *J Pediatr*. 1983;102(1):161.
- Velanovich V. Ectopic breast tissue, supernumerary breasts, and supernumerary nipples. *South Med J*. 1995;88(9):903-906.
- Pryor LS, Lehman JA, Workman MC. Disorders of the female breast in the pediatric age group. *Plast Reconstr Surg*. 2009;124(1):50e-60e.
- Dixon JM, Mansel RE. ABC of breast diseases: congenital problems and aberrations of normal breast development and involution. *BMJ*. 1994;309(6957):797-800.
- Haber HP, Wollmann HA, Ranke MB. Pelvic ultrasonography: early differentiation between isolated premature thelarche and central precocious puberty. *Eur J Pediatr*. 1995;154(3):182-186.
- Lanciotti L, Cofini M, Leonardi A, Penta L, Esposito S. Up-to-date review about minipuberty and overview on hypothalamic-pituitary-gonadal axis activation in fetal and neonatal life. *Front Endocrinol*. 2018;9:410.
- Acer T, Derbent M, Hiçsönmez A. Bloody nipple discharge as a benign, self-limiting disorder in young children: a systematic review including two related case reports. *J Pediatr Surg*. 2015;50(11):1975-1982.
- Lee HB, Roh TS, Chung YK, Kim SW, Kim JB, Shin KS. Correction of inverted nipple using strut reinforcement with deepithelialized triangular flaps. *Plast Reconstr Surg*. 1998;102(4):1253-1258.
- Einav-Bachar R, Phillip M, Aurbach-Klipper Y, Lazar L. Prepubertal gynaecomastia: aetiology, course and outcome. *Clin Endocrinol*. 2004;61(1):55-60.
- Dunbar B, Dündar N, Ecri T, Bober E, Büyükgebiz A. Leptin levels in boys with pubertal gynecomastia. *J Pediatr Endocrinol Metab*. 2005;18(10):929-934.
- Eidlitz-Markus T, Mukamel M, Haimi-Cohen Y, Amir J, Zeharia A. Breast asymmetry during adolescence: physiologic and non-physiologic causes. *Isr Med Assoc J*. 2010;12(4):203-206.
- Chung EM, Cube R, Hall GJ, González C, Stocker JT, Glassman LM. From the archives of the AFIP: breast masses in children and adolescents: radiologic-pathologic correlation. *Radiograph*. 2009;29(3):907-931.
- Kulkarni D, Dixon JM. Congenital abnormalities of the breast. *Womens Health*. 2012;8(1):75-88.
- Sadove AM, van Aalst JA. Congenital and acquired pediatric breast anomalies: a review of 20 years' experience. *Plast Reconstr Surg*. 2005;115(4):1039-1050.
- Wolfswinkel EM, Lemaine V, Weathers WM, Chike-Obi CJ, Xue AS, Heller L. Hyperplastic breast anomalies in the female adolescent breast. *Semin Plast Surg*. 2013;27(1):49-55.
- Warren R, Degnim AC. Uncommon benign breast abnormalities in adolescents. *Semin Plast Surg*. 2013;27(1):26-28.
- Restrepo R, Cervantes LF, Swirsky AM, Diaz A. Breast development in pediatric patients from birth to puberty: physiology, pathology and imaging correlation. *Pediatr Radiol*. 2021;51(11):1959-1969.