

## Case Report

# Familial Gynecomastia of Unknown Etiology: A Case Report

Ishant kumar Chaurasia<sup>1</sup>, Fahim Rajiwate<sup>1</sup>, Ashish Jadhav<sup>1</sup>,  
Ashok Kalyanshetti<sup>1</sup>, Mahim Koshariya<sup>2\*</sup>

<sup>1</sup>Department of General Surgery, MGM Hospital, MGM University of Health Sciences, Kamothe, Navi Mumbai, Maharashtra, India

<sup>2</sup>Department of General Surgery, Gandhi Medical College and Associated Hamidia Hospital, Bhopal, Madhya Pradesh, India

\*Corresponding author email: [mahimk2000@yahoo.co.uk](mailto:mahimk2000@yahoo.co.uk)

	International Archives of Integrated Medicine, Vol. 2, Issue 11, November, 2015. Copy right © 2015, IAIM, All Rights Reserved. Available online at <a href="http://iaimjournal.com/">http://iaimjournal.com/</a>	
	ISSN: 2394-0026 (P)	ISSN: 2394-0034 (O)
	Received on: 01-11-2015	Accepted on: 07-11-2015
	Source of support: Nil	Conflict of interest: None declared.
<b>How to cite this article:</b> Chaurasia IK, Rajiwate F, Jadhav A, Kalyanshetti A, Koshariya M. Familial Gynecomastia of Unknown Etiology: A Case Report. IAIM, 2015; 2(11): 67-70.		

## Abstract

Gynecomastia is a benign enlargement of the male breast (usually bilateral but sometimes unilateral) resulting from a proliferation of the glandular component of the breast. It is a common clinical condition. We have reported here a case of 16 years old male who presented with bilateral gynecomastia. In his family history his Grandfather, Father and cousin brother suffered from same disease. As all the relevant investigations came out to be normal, it was considered as idiopathic condition and was diagnosed as familial gynecomastia.

## Key words

Gynecomastia, Unknown etiology, Familial.

## Introduction

Gynecomastia is common and most prevalent in the neonatal, pubertal and elderly periods [1]. Gynecomastia results from proliferation of the glandular breast compartment triggered by several endogenous, and occasionally by exogenous, factors [2]. Often, gynecomastia is the result of absolute imbalance between estrogen and androgen action at the breast tissue

level. Estrogens stimulate and androgens inhibit breast glandular development [2, 3]. Age, family history, drug/medication history, clinical manifestation and specific diagnostic modalities remain crucial in differentiation and treatment. There are various causes of gynecomastia but in many cases, esp. familial, the cause is unknown. Presenting a case of Familial Gynecomastia and reviewing related literature.

## **Case report**

A 16 years old male presented with enlargement of both the breasts since 5 years (**Figure - 1**). Patient was 11 years old when he noticed that both his breasts were gradually increasing in size. There was no history of pain, fever or any kind of discharge from both nipples. Family history of patient revealed that enlargement of bilateral breast in father (**Figure - 2**), grandfather, paternal cousin brother (1<sup>st</sup> degree relative) (**Figure - 3**). Physical examination noted both the breasts were fully developed simulating female breasts. No axillary lymph nodes were palpable. The external genitalia was well developed with secondary sexual characteristics normal for his age. The remainder of the physical exam was unremarkable.

**Figure – 1:** Bilateral Gynecomastia.



Ultrasound reported fibrocystic changes within bilateral breast. Serum testosterone, serum beta-HCG, serum prolactin, serum FSH was done which came normal. Also the chromosomal study showed no structural abnormality.

**Figure – 2:** Bilateral gynecomastia in his father.



**Figure – 3:** Bilateral gynecomastia in his cousin.

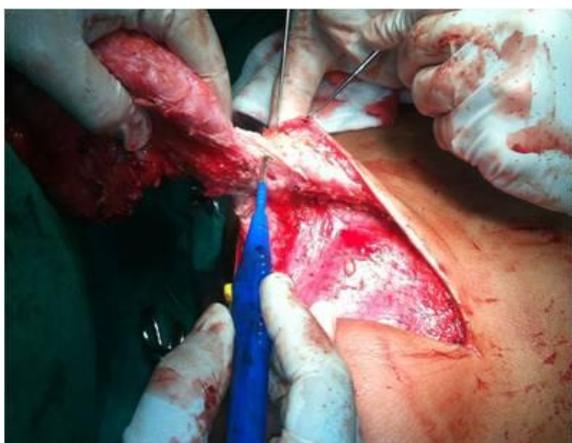


Bilateral mastectomy was done (**Figure - 4, 5, 6**) and specimen was sent for histopathology and ER status. Histopathology report showed ducts lined by cuboidal epithelium with absence of terminal inter ductal unit. ER status of the specimen was positive.

**Figure – 4:** Elevation of flap through inframammary incision.



**Figure – 5:** Excision of lump.



**Figure – 6:** Bilateral mastectomy.



## Discussion

Gynecomastia has been a curious disease since ancient times. There are many examples of historical statues demonstrating gynecomastia. A famous example is that of the 18<sup>th</sup> Dynasty pharaoh Akhenaten. Another example is of a zulu chief, Chengwayo who had forty wives and over 100 children. He had breast as large as a woman's and had nursed some of his children. He was keenly alive to the fact that he was different from common run of men and took great pride in it.

The term "gynecomastia" was introduced in the second century A.D. by the famed Roman physician and philosopher, Galen. Byzantine physician Paulus of Aegina described a surgical procedure employing a "lunated" (moon-shaped) sub-mammary incision with provision for resection of skin in pendulous cases.

It is a benign enlargement of the male breast as a result of proliferation of the glandular component, is common, being present in 30-50% of healthy men [4, 5]. It is caused by an increase in the ratio of oestrogen to androgen activity [6]. The condition may be bilateral or unilateral.

There are innumerable causes of gynecomastia, but in many cases there is no possible cause found as it was in the case described above. In this case patient and all the affected family members had well developed genitals. As his father and grandfather have produced offspring, there was no question about the functions of their reproductive organs. His cousin brother and the patient himself had normal Sexual maturity rating (Maxwell and Tanner). All relevant blood investigation like Serum testosterone, serum beta-HCG, serum prolactin, serum FSH, along with chromosomal study done for the patient was normal. So, the only two possibilities which could be considered as etiologic factor in this case were either aromatase excess or increased sensitivity /increased receptors of breast tissue for estrogen.

Aromatase excess syndrome, seen in familial gynecomastia, is a rare genetic and endocrine syndrome, which is characterized by over-expression of aromatase enzyme responsible for conversion of androgen to estrogen. It is inheritable, autosomal dominant genetic mutation affecting CYP19A1 gene which is located on short arm of 15th chromosome (15q21.1). As the facility of determining the level of aromatase enzyme was not available, this test could not be done.

Increased sensitivity of the breast tissue was evaluated by doing estrogen receptor (ER) status of the histopathology specimen of breast, which came out to be positive.

### **Conclusion**

The Gynecomastia has been a curious disease since ancient times. Many theories have been proposed but not a single dependable argument holds good in many unilateral cases of gynecomastia and the curiosity remains. We have here reported a case of familial gynecomastia of unknown etiology because of its rarity as only

few cases are reported in available medical literature.

### **References**

1. Niewoehner C, Schorer AB. Gynaecomastia and breast cancer in men. *BMJ*, 2008; 336: 709-13.
2. Braunstein G.D. Male Reproductive Endocrinology. *Cecil's Essentials of Medicine*. Edited by Thomas E. Andreoli, et al., WB Saunders, 2001, p. 575-582.
3. Wilson J.D. Endocrine Disorders of the Breast. *Harrison's Principles of Internal Medicine*. Edited by Kurt Isselbacher, et al., NewYork: McGraw-Hill, 1997, p. 111-115.
4. Nuttall FQ. Gynaecomastia as a physical finding in normal men. *J Clin Endocrinol Metab.*, 1979; 48: 338-40.
5. Niewoehner CB, Nuttall FQ. Gynaecomastia in a hospitalized male population. *Am J Med.*, 1984; 77: 633-8.
6. Braunstein GD. Gynaecomastia. *N Engl J Med.*, 1993; 328: 490-5.