

Congenital Malformations and Developmental Changes of the Breast: A Neonatological View

P. Merlob

Department of Neonatology, Rabin Medical Center, Beilinson Campus, Petah Tiqva and Sackler Faculty of Medicine, Tel Aviv University, Tel Aviv, Israel

ABSTRACT

Congenital malformations of the breast and nipple-areolar complex are described from the aspect of neonatal dysmorphology. Their classification is based on the accepted nomenclature in the medical literature and on a special method of clinical examination. The embryology and the possible genes involved in breast development are also discussed to clarify the mechanisms underlying the occurrence of breast malformations. Except for amastia, polymastia and polythelia, which have received much attention, the data for many other breast malformations are sparse. Case control studies or even case series are rare, and good evidence is often lacking. In addition, more studies are needed to complete the map of genes involved in breast development. This information will contribute substantially to our understanding and clinical approach to breast malformations in the future.

KEY WORDS

amastia, polymastia, athelia, polythelia, breast and nipple malformations

I. INTRODUCTION

A congenital malformation is a morphologic defect (error of morphogenesis) of an organ, part of an organ, or larger region of the body that is present at the time of birth and is the result of an intrinsically abnormal developmental process^{1,2}. That is, the abnormality is present already in the primordium, or the group of cells in the embryo that represents the first trace of an organ, so that the potential for development of a normal form or structure is nil to begin with.

In the breast, congenital malformations may involve the breast gland or the nipple-areolar complex. Though already existing at birth, some may become visibly apparent only in puberty. The classification of breast malformations used in this review (see Table 1) was based on the accepted nomenclature in the medical literature^{2,3} and on the method of examination used in our neonatal department, called the PPSSAC system ('psac' means decision in Hebrew), wherein every organ in the body is rated for six features: presence, position, size, shape, adjacent area and components.

II. BREAST EMBRYOLOGY AND DEVELOPMENT

To understand the occurrence of congenital breast malformations, an understanding of breast embryology is necessary.

The mammary glands are highly modified sweat glands that originate from surface epithelium growing down into the underlying mesenchyme⁴. Breast embryogenesis takes place in several stages, though the timing (in weeks gestation) is relatively flexible, and some of the stages may overlap (Fig. 1).

Reprint address:
P. Merlob, M.D.
Department of Neonatology
Rabin Medical Center
Beilinson Campus
Petah Tiqva 49100, Israel
e-mail: merlobp@post.tau.ac.il