Genetic Abnormalities of Primary Amenorrhea in Women from the South of Iraq

Saad A. Al Omar [1], Zinah Anas Salman [2], Ahmed Kamal Kasim [3]

1. Department of Pathology, College of Medicine, University of Basrah, Basrah, Iraq.
2. Department of Pediatrics, Maternity and Pediatric General Hospital, Basrah Health Directorate.
3. Department of Cytogenetics, Baghdad Teaching Laboratories, Medical City, Baghdad.

Received: 2.1.2024 **Accepted: 16.1.2024**

Abstract:

Background: Genes and hormones play an important and complicated role in differentiating the gonads to the testis or ovary. Primary or secondary amenorrhea is either absence or unexpected early end of menstruation. Hormonal, physiological, environmental and genetic reasons are all involved in developing such a disorder. Diagnosis is very important to provide the required treatment and one of the powerful diagnostic goals is Karyotyping.

Aim of study: So, the study aimed to identify the chromosomal abnormalities through karyotyping and how frequent each is.

Methods: Samples of 174 patients who were referred to AlBayan Private Laboratory in Basrah City from 2018 to 2022

Results: A total of 174 patients were diagnosed with PA and the results showed that out of 174 patients, 57(30%) were diagnosed with chromosomal abnormalities using cytogenetics. Karyotype analysis showed that 46, XY represented more than 50% of the genetic abnormalities, followed by 27% and 3% due to 45, X or 46, X, i(Xq) respectively. However, all the rest abnormalities displayed the same percentage, 1% of the total number of PA patients. Our study showed that a significant number of cases with primary amenorrhoea harbor chromosomal abnormalities, which are significant in gonadal dysgenesis.

Conclusion: So genetic counselling, routine chromosomal study, hormonal assessment, and radiological evaluation are important for proper management, also the hormonal replacement for Turner Syndrome patients, and screening for malignancy in patients with sex reversal are important

Keywords: primary amenorrhea, karyotyping, chromosomal abnormalities, congenital anomalies

Corresponding author: Saad A. Al Omar

College of Medicine, University of Basrah, Basrah, Iraq.

✉ **E-mail:** saad.abdullah@uobasrah.edu.iq

Introduction

Normally, the menstrual function is expected at puberty, but if this doesn't happen then it is a disease called primary amenorrhea (PA) (1), which is defined as no pubertal signs, by the age of 13 years or 5 years after breast development and no menarche occurs. When the menstruation ceases after regular cycles this is called secondary amenorrhea (2). Karyotype analysis shows that 3.4% of PA patients are XY females. Despite that the incidence of PA doesn't exceed 5%, the incidence is increasing due to better registration, social media and better health care (3). One of the main reasons for PA is the congenital anomalies including Mullerian aplasia or Mayer-Rokitansky-Kuster Hauser Syndrome (MRKHS) (4). This involves the absence of the uterus and vagina in addition to obstruction of the reproductive tract. Gonadal dysgenesis could be considered as another reason for PA (5). Hypothalamic disorders are also counted as one of the main reasons which could be represented as a gonadal or/and ovarian disorder (6). One of the diagnostic keys of PA is karyotyping to identify the genetic abnormalities underlying this disorder with various percentages of each chromosomal defect (7). So, the study aimed to identify the chromosomal abnormalities, how frequent each is in patients and the value of knowing the genetic reason to treat accordingly.

10.33762/mjbu.2024.145705.1183