MEDWIN PUBLISHERS
Committed to Create Value for Researchers

Journal of Human Anatomy
ISSN: 2578-5079

Status on the Intra-Chromosomal Aberrations with Some Sorts of Hormonal Disturbances in Human

Kabir A*

Department of Biology, Cantonment Public College, Bangladesh

***Corresponding author:** Ashraful Kabir, Department of Biology, Cantonment Public College, Saidpur Cantonment-5311, Nilphamari, Bangladesh, Email: ashraful.mission@gmail.com

Mini Review

Volume 8 Issue 1

Received Date: December 12, 2023

Published Date: January 31, 2024

DOI: 10.23880/jhua-16000191

Abstract

Chromosomal aberration due to wrong number or for some hormonal imbalances, humans show chromosomal anomalies at birth or in adolescence. Those are fatal for entire human life. Many patients cannot survive, and morbidity may happen later. An article helped to write this qualitative review of chromosomal abnormalities. In this aspect, if we know the true causes of various chromosomal and hormonal imbalances, we could get solution. Only motivation and knowledge on this issue could help to eradicate these problems from the society at all.

Keywords: Chromosomal Aberration; Hormonal Disturbances; Human; Causes; Prevention

Introduction

Clinical cytogenetics began its rapid advancement with the discovery of the correct chromosome numbers (46) in human in 1956 [1,2], then various types of major chromosomal syndromes with modified numbers of chromosomes such as Down syndrome (trisomy 21), Turner's syndrome (45, X) and Klinefelter's syndrome (47, XXY) were detected [2]. About 1000 chromosomal abnormalities have been identified to this time. This made a major contribution to human morbidity and mortality [3]. The common chromosomal abnormalities of human are mental retardation, congenital malformation, sterility, sexual abnormalities and spontaneous fetal loss [4]. Congenital abnormalities with chromosomal defects cause gross phenotypic anomalies that lead to mental retardation [5,6]. The disorders of sexual development associated with abnormal karyotype of sex chromosome and Turner's syndrome, Klinefelter's syndrome, certain menstrual disorders (primary and secondary amenorrhea), superman syndrome, true and pseudohermaphroditisms [1]. 37.5% individuals showed pericentric inversions with Down syndrome, intellectual disability, dysmorphic features, congenital anomalies, developmental delay, Turner's syndrome, and Klinefelter's syndrome [1]. Risk of unbalanced gametes depends on the location of chromosome break points relative to the centromere and cross-over frequency [7]. Infertility and recurrent abortion as it can act on acentric fragments formed by meiosis and synaptical complexes [8]. The cause of 21 trisomy (Down syndrome) is the most frequent aneuploidy in human populations and found in newborn with 1:700 [9]. The majority of fetuses with Edwards' syndrome and spontaneously aborted, it is not common to patients [10,11]. Turner's syndrome afflicts approximately 1 in 2000 females and it is most common factor in infertile women [12]. The second frequency of sex chromosomal aberrations was Klinefelter's syndrome being more frequent in infertile males [12]. The objective of this study is to understand the causes of chromosomal aberrations with some hormonal disturbances due to take preventive measures against such problems (Table 1).

Name of Problems	Causes	Chromosome Number	Salient Features
True hermaphrodite	Tetrasomy (XXXY)	48	Testicular and ovarian tissue are present; sexually active but sterile
Pseudofemale (female intersex)	Secondary sexual characteristics (XX)	46	Female-like appearance but not completely female
Pseudomale (male intersex)	Secondary sexual characteristics (XY)	46	Male-like appearance but not fully male
Wolf-Hirschhorn syndrome	Small deletion from 4 pairs of chromosomes	46	Low muscle tone (hypotonia); IgA deficiency; congenital heart defect
Cri-du-chat	Small deletion from 5 pairs of chromosomes	46	Cat-like crying
Down's syndrome	21 trisomy but Robertsonian translocation happens between 14 and 21 pair of chromosomes	46	Eye distance is large
Triploid syndrome	Euploidy (3n or 4n)	69 or 92	Spontaneous abortion; stillbirth; neonatal death
Patau's syndrome	13 trisomy	47	Cleft lip; polydactyly; abnormal genitalia; congenital cardiac defect; various defective organs
Edwards' syndrome	18 trisomy	47	Deformed skeletal system; congenital cardiac defect; defective kidney, colon, and small intestine; deformed ear
Turner's syndrome	Monosomy (XO)	45	Sterile; narrow pelvis; underdeveloped breasts
Triplo-x female/ Superfemale	Trisomy (XXX)	47	Female but with infantile genitalia at maturity (some females are found in mental hospital)
Supermale	Trisomy (XYY)	47	Over 6 feet tall; low IQ; commit crime
Klinefelter's syndrome	Trisomy (XXY)	47	Enlarge breasts; broad pelvis; sterile; small testes; criminally insane

Table 1: All about humans chromosomal and hormonal disturbances [13].

Conclusion

Consanguineous marriages culturally accomplish in most Asian and African populations especially in Muslim countries. It is apparent that these types of marriage are an influential element for some autosomal recessive genetic disorders [14]. A study clearly demonstrated twelve types of genetic disorders in the case of cousin marriages [15]. Especial motivation to poor people is mandatory. In our education syllabus, we could implement a chapter on chromosomal aberration in humans. These chromosomal aberrations are few, to make people conscious to minimize these genetic disasters in the society. An accurate laboratory diagnosis using florescence in-situ hybridization and other complementary molecular approaches is needed [1].

References

1. Ghazaey S, Mirzaei F, Ahadian M, Keifi F, Tootian S, et al. (2013) Pattern of chromosomal aberrations in patients from North East Iran. Cell Journal 15(3): 258-265.

2. Smeets DF (2004) Historical prospective of human cytogenetics: from microscope to microarray. Clinical Biochemistry 37(6): 439-446.

3. Goud MT, Al-Harassi SM, Al-Khalili SA, Al-Salmani KK, Al-Busaidy SM, et al. (2005) Incidence of chromosome abnormalities in the Sultanate of Oman. Saudi Medical Journal 26(12): 1951-1957.

4. Shah VC, Murthy DS, Murthey SK (1990) Cytogenetic studies in a population suspected to have chromosomal abnormalities. Indian Journal of Pediatrics 57(2): 235-243.

5. Pashayan H, Dallaire L, MacLeod P (1973) Bilateral aniridia, multiple webs and severe mental retardation in a 47, XXY-48, XXXY mosaic. Clinical Genetics 4(2): 125-129.

6. Kour A, Mahajan S, Singh JR (2003) Cytogenetic profile of individuals with mental retardation. International Journal of Human Genetics 3(1): 13-16.

7. Luthardt FW, Keitges E (2001) Chromosomal syndromes and genetic disease. Encyclopedia of life sciences, Nature Publishing Group.

8. Mozdarani H, Mohseni Beybodi A, Zari-Moradi S (2008) A cytogenetic study of couples with recurrent spontaneous abortions and infertile patients with recurrent IVF/ICSI failure. Indian Journal of Human Genetics 14(1): 1-6.

9. Dursun P, Gultekin M, Yuce K, Ayhan A (2006) What is the underlying cause of aneuploidy associated with increasing maternal age? Medical Hypotheses 66(1): 143-147.

10. Duarte AC, Cunha E, Roth JM, Ferreira FL, Garcias GL, et al. (2004) Cytogenetics of genetic counseling patients in Pelotas, Rio Grande do Sul, Brazil. Genetics and Molecular Research 3(3): 303-308.

11. Giaccardi A, Sardi R, Priora U, Vivalda M, Domeneghetti G, et al. (1991) Trisomy 18 or Edwards' syndrome. A report of 4 clinical causes. Minerva Pediatrics 43: 343-349.

12. Salahshourifar I, Sadat Masoudi N, Gourabi H (2009) Cytogenetic findings in couples who are candidates for assisted reproductive techniques. Yakhteh 10(4): 288-294.

13. Islam MS (2018) *Selected Lectures on Genetics.* Lambart Academic Publishing, Germany, pp: 300.

14. Hasanzadeh-Nazarabadi M, Rezaeetalab GH, Dastfan F (2006) Study of youths' knowledge, behavior, and attitude towards consanguineous marriages. Iranian Journal of Public Health 35(3): 47-53.

15. Kabir A (2019) Comparative studies on birth disorders in cousin and unrelated marriages. Scientific Periodique Medicine 7(6): 1-11.