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Original Research Article

Ten-year experience retrospective in investigation of the frequency of sex chromosome aneuploidies diagnosed in a reference medical center in Türkiye

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ABSTRACT

Background: Sex chromosome aneuploidies (SCAs) are characterized by the gain or loss of entire or parts of sex chromosomes.
Methods: In 5212 cases studied over the last 10 years in Medical Genetics Department, we found 3793 (72.78%) other chromosomal establishments and 1419 (27.22%) sex chromosomal aneuploidies using the retrospective archive-file scanning method.
Results: 839 (31.32%) of the 1419 cases with anomalies were female, and 580 (22.8%) were male. While Triple X-Super Female Syndrome 423 (15.8%) is the most common in women, Klinefelter Syndrome 508 (20.0%) is the most common in men. Among the cases sent to our outpatient clinic for various indications (prediagnosis) for karyotype analysis, 1447 (54.0%) in women and 1193 (47.1%) in men were the cases with a prediagnosis of habitual abortion; followed by 298 (11.1%) infertility in women and 350 (13.8%) in men. While 167 (6.6%) people were sent with the preliminary diagnosis of azoospermia in men, it was determined to be 124 (4.6%) in women with primary amenorrhea. In the analysis we made according to the diagnosis, 1839 (68.7%) women and 1954 (77.1%) men were found to be normal. While the most important diagnosis is Triple X-Super Female syndrome 423 (15.8%) in women, Klinefelter syndrome 508 (20.0%) is the first in men. Turner Syndrome is found in 315 (11.8%) women, while Jacob's syndrome (47, XYY) is detected in 13 (0.5%) men.
Conclusions: As a result, the retrospective evaluation of 10-year cytogenetic analyses of these case groups contributes to the creation of regional data.

Keywords: Chromosomal anomalies, Sex developmental disorders, Cytogenetic analysis

INTRODUCTION

Sex chromosome aneuploidies (SCAs) are the most commonly occurring chromosomal disorders, affecting 1 in 400 newborns, and are an important health problem due to the material and moral difficulties they bring to the patient's family and society as well as to the patient[1]. All the SCAs are characterized by the gain or loss of all (aneuploidy) or parts of a part (structural abnormalities, e.g., isochromosomes) of the sex chromosomes, and among the SCAs, the best-known syndromes are Turner syndrome (45, X), Klinefelter syndrome (47, XXY), Triple X syndrome (47, XXX) and Jacob's syndrome (47, XYY).[2] Among these malformations, gender development irregularities constitute an important subgroup. The sex chromosomes, X and Y chromosomes, are primarily responsible for determining the sex, but aneuploidies occurring in these chromosomes can lead to

syndromes that are important enough to change the sociocultural development of the child to be born. In addition, the detection of 47, XXX, and 45, X anomalies in females and showing that 47, XXY individuals are males emphasizes the importance of the sex-determining role of the human Y chromosome.[3] Clarifying the etiology of the disorder in children is extremely important in terms of giving healthy genetic counselling to families. Sex chromosome aneuploidies (SCAs) are a common family of genetic disorders that collectively increase the risk of neuropsychiatric and cognitive impairment. SCAs are more than important medical disorders in their own right; they offer a unique naturally occurring model for studying X and Y chromosome effects on the human brain.[4]

Human sex chromosome aneuploidies are noteworthy in several different ways. First, their mechanism is unique according to their maternal or paternal origin: unlike all autosomal aneuploidies, sex chromosome aneuploidies typically result from the loss or gain of a paternal chromosome. Second, the phenotypic traits observed in individuals with a sex abnormality are milder compared to autosomal imbalance, and fertility is still possible for some individuals. Third, despite abnormal sex chromosome structures, most autosomal chromosomes are normal.[3] Two mechanisms have still not lost their importance in the occurrence of possible aneuploidies; anaphase lagging and nondisjunction. Particularly before or during meiosis, the loss or increase of all or part of the sex chromosomes are division a defect that occurs with a significant frequency.

Sex chromosome abnormalities, as with autosomal chromosomes, can be numerical as well as structural, present in whole cells or mosaic form. We detected mosaic forms mostly as mixed gonadal dysgenesis. According to the results of the study for Klinefelter Syndrome, approximately 50% of cases are due to nondisjunction in paternal meiosis I. This is the only possible paternal origin of 47, XXY cases, as errors in paternal meiosis II will produce XX or YY gametes.[5] 47, XXX is found in 1 out of every 1000 female births.[6] The 47, XYY (Jacob's Syndrome) condition is unique because it necessarily involves a paternal error.

Only Y chromosome nondisjunction occurring during paternal meiosis II or during the early cleavage divisions can produce a conceptus with two Y-chromosomes.[7] 47, XYY occurs in 1 in 1000 males.[8] At 47, XYY results from the production of a YY sperm by nondisjunction at the second paternal meiotic division or from postfertilization nondisjunction of the Y chromosome. 45, X is a common chromosomal disorder that affects approximately 1 in 2500 to 1 in 2000 live-born female infants and is characterized by short stature and a mane neck, and the X chromosome is usually of maternal origin. [9,10] Overall, 50% of patients have 45, X, 17% have an isochromosome of the long arm of X, 24% are mosaics, 7% have a ring X, and 2% have a short arm deletion of one X. [11]

The incidences of live-born children, fetuses examined in the prenatal period, and spontaneous abortions are important subject of investigation. The use of cytogenetics for the examination of sex chromosome abnormalities is considered the gold standard in Medical Genetics. Therefore, this study aimed to evaluate and discuss the cytogenetic results of our found cases of sex chromosome abnormalities (1419 of 5212 cases).

METHODS

Cytogenetic method and analysis

The study was conducted retrospectively using the cytogenetic findings from 5212 cases obtained between 2011-2021 and were reviewed at Afyonkarahisar Health Sciences University Faculty of Medicine, Department of Medical Genetics. Chromosomes were obtained by working with heparinized blood samples according to the standard procedure.

In the said standard procedure, the steps of the operation are, respectively, peripheral venous blood samples taken into heparin tubes for chromosome analysis, inoculated into RPMI-1640 medium with phytohemagglutinin (PHA), and cultured at 37ºC for 72 hours. Colcemid is added 45 minutes before the study. Cultured blood cells are lysis with the hypotonic solution and also fixed with Carnoy's fixative. The cell suspension is spread on the slides and the aging process is applied. Obtained chromosomes are analyzed with at least 25 metaphase plates after GTG banding. The cytogenetic analyses of the patients were studied in accordance with the ISCN (2009, 2013, and 2016) protocols.

Inclusion criteria

From the patient files that were studied, analyzed and archived with the standard chromosome acquisition method, those with sex chromosome anomalies were selected. Investigated patients were grouped according to clinical features and reasons for admission. Chromosome analysis results and follow-up records of people who were diagnosed with sex chromosome anomalies after completing the cytogenetic analysis were included in the study.

Exclusion criteria

While retrospectively scanning the patient records in the cytogenetics unit of our Medical Genetics department, patient files containing the analysis results of autosomal chromosomes, excluding gender anomalies, were not included in the study.

Statistical analysis

The numbers and percentages were given as descriptive statistics. Pearson A Chi-Square test was used to evaluate whether there was a difference between genders in terms of years, diagnoses, and indications. A $p<0.05$ value was determined as the level of significance. The SPSS 20.0 package program was used in the analysis. Since our study is not a sample study, it is not included because it is not necessary to calculate the sample size. Our study is a single-center population study conducted with a 10-year retrospective archive scanning method.

RESULTS

Sex chromosome anomalies are the most frequently observed chromosomal anomalies in prenatal and postnatal diagnosis, and their incidence is 1 in 448 newborns.[12] In order to determine the etiology in this case group, phenotypic findings were analyzed by correlating them with cytogenetics. Example karyotypes of common sex chromosome anomalies obtained in our laboratory are given in (Figure 1).

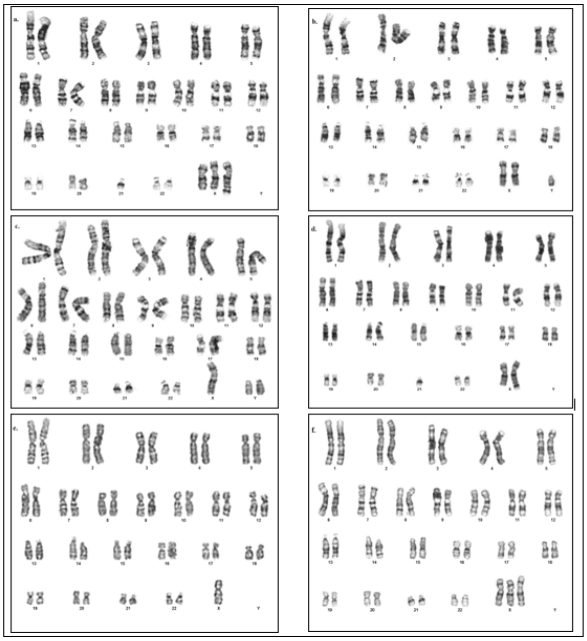

Figure 1: Sample karyotypes obtained in our cytogenetic laboratory of our patients with sex chromosome anomaly a. 47,XXX,t(X;21) b.47,XXY c.47,XYY d.46,XX,t(X;21) e.45,X f.47,XXX.

In our study, the distribution of sex chromosome anomalies according to years was evaluated in cases that applied to our clinic with the preliminary diagnoses given in (Table 1). Retrospective evaluation of 10-year cytogenetic analyses of these case groups contributed to the creation of regional data. In the evaluation made according to years, our cases who applied to our clinic with the prediagnosis of habitual abortion, azoospermia, and infertility constitute the first three lines. As in similar publications, the most common admissions to our clinic are cases with intellectual disability, dysmorphic features, congenital anomalies, developmental delay, abnormal karyotypes, amenorrhea, male infertility, ambiguous genitalia, intellectual disability, dysmorphic features, congenital anomalies, and developmental delay, and recurrent miscarriage.[13] In our study, 5212 cases were screened, 3793 (72.78%) cases were found to be normal, and 1419 (27.22%) sex chromosome anomalies were detected. Duarte *et al.*, Singh, and Santos *et al*, have reported a higher frequency of chromosome abnormalities in their investigations (29.3%, 28.8%, and 28.6%, respectively).[14–16] Of 1419 cases with a sex chromosome anomaly, 839 (31.32%) were female and 580 (22.8%) were male. Our culture success rate was 98.4%. Al-Alawi et al. in their study, as in our study, supported the importance of cytogenetic analysis in patients with pre-diagnoses of ambiguous genitalia, turner syndrome, klinefelter syndrome, and amenorrhea and found to have sex chromosome anomaly.[12] Among the cases sent to our outpatient clinic with various prediagnoses for karyotype analysis, 1447 (54.0%) in women and 1193 (47.1%) in men were the cases with a prediagnosis of habitual abortion, 298 (11.1%) in women and 350 (13.8%) in men were the cases followed by infertility. 232 (8.7%) women and 213 (8.4%) men were sent with the suspicion of other genetic syndromes. While 167 (6.6%) people were sent with the prediagnosis of azoospermia in men, it was determined to be 124 (4.6%) in women with Primary Amenorrhea (Table 2). Although Down syndrome is not a sex chromosome anomaly, a sex chromosome anomaly was detected in 11 (6.9%) of 180 cases with this prediagnosis. Different sex chromosome anomalies were found in 86 (3.2%) women and 94 (3.7%) men who applied with the prediagnosis of Down syndrome. In the analysis we made according to the diagnosis; 1839 (68.7%) women and 1954 (77.1%) men were found to be normal. While the most important diagnosis is Triple X-Super Female syndrome 423 (15.8%) in women, Klinefelter syndrome 508 (20.0%) is the first in men. Turner syndrome is found in 315 (11.8%) women, while Jacob's syndrome (47, XXY) and 13 (0.5%) in men were detected (Table 3). Duarte et al. (2004) reported that Klinefelter syndrome was the most common sexual abnormality and only 20% of cases showed a classic, well-defined phenotype, which is similar to our study. [14] In our study, while 855 (59.1%) of 1147 cases in women with a prediagnosis of habitual abortion were normal, 217 (15%) of Turner syndrome and 375 (25.9%) of Triple-X were diagnosed (Table 4). In our study, 154 (92.2%) of the cases who applied with the prediagnosis of azoospermia in men were diagnosed with Klinefelter syndrome (Table 5). Considering other studies, our study strongly supports the relationship between prediagnosis of azoospermia and Klinefelter syndrome.[17] Chromosomal abnormalities are one of the most important causes of male infertility. Although Klinefelter syndrome patients are considered infertile as a general opinion, fertilization can be achieved with some assisted reproductive techniques.

Table 1: The distribution of sex chromosome anomalies according to years was evaluated in cases that applied to our clinic with the preliminary diagnoses.

Indıcatıon	Gender	2021	2020	2019	2018	2017	2016	2015	2014	2013	2012	2011
Habitual abortion in his wife	Female	96 (47.8)	84 (0.9)	139 (51.7)	148 (52.5)	170 (57.4)	205 (52.3)	141 (57.1)	131 (58.7)	121 (58.7)	156 (53.1)	56 (52.8)
	Male	94 (41.8)	68 (46.3)	114 (52.1)	124 (43.5)	135 (47.7)	147 (36.4)	104 (46.4)	102 (47.0)	132 (52.0)	132 (52.0)	41 (56.9)
Azoospermia	Female	-	-	-	-	-	-	-	-	-	-	-
	Male	21 (9.3)	20 (13.6)	26 (11.9)	34 (11.9)	31 (11.0)	20 (5.0)	13 (5.8)	2 (0.9)	-	-	-
Infertility	Female	24 (11.9)	9 (5.6)	16 (5.9)	37 (13.1)	32 (10.8)	43 (11.0)	32 (13.0)	37 (16.6)	34 (15.5)	32 (10.9)	4 (3.8)
	Male	27 (12.0)	11 (7.5)	14 (6.4)	42 (14.7)	26 (9.2)	55 (13.6)	38 (17.0)	55 (25.3)	36 (17.6)	36 (14.2)	10 (13.9)
Down syndrome	Female	10 (5.0)	6 (3.7)	13 (4.8)	10 (3.5)	8 (2.7)	11 (2.8)	9 (3.6)	7 (3.1)	4 (1.4)	4 (1.4)	4 (3.8)
	Male	18 (8.0)	6 (4.1)	11 (5.0)	10 (3.5)	12 (4.2)	12 (3.0)	6 (2.7)	13 (6.0)	-	-	6 (8.3)
Genetic syndromes	Female	5 (2.5)	6 (3.7)	13 (4.8)	13 (4.6)	18 (6.1)	4 (1.0)	4 (1.6)	4 (1.8)	14 (6.8)	16 (5.4)	1 (0.9)
	Male	11 (4.9)	9 (6.1)	11 (5.0)	12 (4.2)	20 (7.1)	7 (1.7)	-	8 (3.7)	13 (6.4)	21 (8.3)	1 (1.4)
Amenorrhea	Female	9 (4.5)	8 (4.9)	14 (5.2)	12 (4.3)	11 (3.7)	13 (3.3)	6 (2.4)	8 (3.6)	6 (2.9)	17 (5.8)	20 (18.9)
	Male	-	-	-	-	-	-	-	-	-	-	-
Hypogonadotropic hypogonaasdism	Female	3 (1.5)	-	1 (0.4)	4 (1.4)	1 (0.3)	-	-	2 (0.9)	-	-	-
	Male	9 (4.0)	1 (0.7)	2 (0.9)	-	-	-	-	-	-	-	-
Developmental retardation	Female	7 (3.5)	8 (4.9)	14 (5.2)	5 (1.8)	5 (1.7)	16 (4.15)	1 (0.4)	4 (1.8)	-	-	-
	Male	7 (3.1)	1 (0.7)	6 (2.7)	4 (1.4)	6 (2.1)	16 (4.0)	7 (3.1)	5 (2.3)	-	-	-
Turner syndrome	Female	8 (4.0)	2 (1.2)	2 (0.7)	5 (1.8)	1 (0.3)	3 (0.8)	-	1 (0.4)	3 (1.5)	6 (2.0)	2 (1.9)
	Male	3 (1.3)	1 (0.7)	1 (0.5)	-	-	-	-	-	-	-	-
Ivf failure	Female	9 (4.5)	18 (11.1)	13 (4.8)	13 (4.6)	15 (5.1)	16 (4.1)	10 (4.0)	1 (0.4)	2 (1.0)	4 (1.4)	10 (9.4)
	Male	6 (2.7)	19 (12.9)	10 (4.6)	12 (4.2)	16 (5.7)	14 (3.5)	10 (4.5)	1 (0.5)	-	6 (2.4)	9 (12.5)
Klinefelter syndrome	Female	-	-	-	-	-	-	-	-	-	-	-
	Male	1 (0.4)	1 (0.7)	2 (0.9)	2 (0.7)	-	-	2 (0.9)	-	-	1 (0.4)	-
Other	Female	23 (11.4)	13 (8.0)	27 (10.0)	19 (6.7)	23 (7.8)	32 (8.2)	20 (8.1)	9 (4.0)	16 (7.8)	43 (14.6)	7 (6.6)
	Male	16 (7.1)	7 (4.8)	14 (6.4)	28 (9.8)	23 (8.1)	54 (13.4)	10 (4.5)	10 (4.6)	14 (6.9)	32 (12.6)	5 (6.9)
Swayer syndrome	Female	-	1 (0.6)	-	-	-	-	-	-	-	-	-
	Male	-	-	-	-	-	-	-	-	-	-	-
Child with anomaly	Female	4 (2.0)	4 (2.5)	8 (3.0)	4 (1.4)	5 (1.7)	7 (1.8)	-	9 (4.0)	5 (2.4)	8 (2.7)	1 (0.9)
	Male	6 (2.7)	3 (2.0)	5 (2.3)	5 (1.8)	7 (2.5)	6 (1.5)	-	8 (3.7)	5 (2.5)	4 (1.6)	1 (0.9)
Neuromotor retardation	Female	1 (0.5)	1 (0.6)	5 (1.9)	5 (1.8)	1 (0.3)	5 (1.3)	5 (2.0)	4 (1.8)	-	2 (0.7)	-
	Male	1 (0.4)	-	2 (0.9)	2 (0.7)	2 (0.7)	3 (0.7)	6 (2.7)	1 (0.5)	-	6 (2.4)	-
Ambiguous genitale	Female	-	1 (0.6)	2 (0.7)	-	-	2 (0.5)	2 (0.8)	2 (0.9)	-	-	-
	Male	1 (0.4)	-	1 (0.5)	-	1 (0.4)	-	1 (0.4)	3 (1.4)	-	6 (2.4)	-
Mental retardation	Female	2 (1.0)	1 (0.6)	2 (0.7)	7 (2.5)	6 (2.0)	35 (8.9)	17 (6.9)	4 (1.8)	3 (1.5)	6 (2.0)	1 (0.9)
	Male	4 (1.8)	-	-	10 (3.5)	4 (1.4)	70 (17.3)	27 (12.1)	9 (4.1)	4 (2.0)	10 (3.9)	-
Total		426	309	488	567	579	796	471	440	412	548	179

Table 2: Among the cases sent to our outpatient clinic with various prediagnoses for karyotype analysis, were the cases with a prediagnosis of habitual abortion, followed by infertility. Among the pre-diagnoses, men were sent with the pre-diagnosis of azoospermia, while primary amenorrhea was found in women.

Prediction	Female, N (%)	Prediction	Male, N (%)
Habitual abortus	1447 (54.0)	**Habitual abortus**	1193 (47.1)
Infertility	298 (11.1)	**Infertility**	350 (13.8)
Other syndromes	232 (8.7)	**Other syndromes**	213 (8.4)
Amenorrhea	124 (4.6)	**Azoospermia**	167 (6.6)
IVF failure	111 (4.1)	**Mental retardation**	138 (5.4)
Genetic syndrome	98 (3.7)	**Genetic syndrome**	113 (4.5)
Down syndrome	86 (3.2)	**IVF Failure**	103 (4.1)
Mental retardation	84 (3.1)	**Down syndrome**	94 (3.7)
Growth retardation	60 (2.2)	**Growth retardation**	52 (2.1)
History of child with anomaly	55 (2.1)	**History of child with anomaly**	49 (1.9)
Turner syndrome	33 (1.2)	**Neuromotor retardation**	23 (0.9)
Neuromotor retardation	29 (1.1)	**Ambiguous genitalia**	13 (0.5)
Hipogonadotropik hipo.	11 (0.4)	**Hipogonadotropik hipo.**	12 (0.5)
Ambiguous genitalia	9 (0.3)	**Klinefelter syndrome**	9 (0.4)
Swayer syndrome	1 (0.0)	**Turner syndrome**	5 (0.2)
Total	2678	**Total**	2534

Table 3: Diagnosis table for men and women.

Diagnosis	Female, N (%)	Diagnosis	Male, N (%)
Normal	1839 (68.7)	**Normal**	1954 (77.1)
Triple- X	423 (15.8)	**Klinefelter syndrome**	508 (20.0)
Turner syndrome	315 (11.8)	**Mix gonodal disgenesis**	26 (1.0)
46,XX,del(X)	37 (1.4)	**46.XY.Yqh+**	19 (0.7)
46,X,i(Xq)	32 (1.2)	**46. X. del (Y) (q11)/45.X**	14 (0.6)
45,X/46,Xr(X)	14 (0.5)	**Jacob syndrome**	13 (0.5)
46,X,tder(X)t(X:21)(q28:q21)mat	10 (0.4)	-	-
Swayer syndrome	8 (0.3)	-	-
Total	2678	**Total**	2534

DISCUSSION

In children with sexual development disorders at later ages; depression, insecurity, feeling lonely, social adjustment problems, rebellion and rejection of family or treatment, self-confidence, behavior problems, self-injury, and even suicide can be seen. Slijper et al found a 39% rate of mental illness in 59 children with CGA.[18]

Therefore, a treatment plan should be established immediately after birth. It is very difficult to answer the question of what constitues "optimal gender selection" in treatment.

The most appropriate sex should be selected by considering the results of the surgical intervention, possible sexual life and reproductive function in the future, after a detailed evaluation by a team consisting of a pediatric endocrinologist, geneticist, pediatric surgeon, experienced psychiatrists and psychologists.

In this process, psychotherapy is recommended in order to clarify gender identity and reveal it to the environment, the process of acceptance, increase social support, and cope with internalized fear.

Group psychotherapy applied to individuals with gender anomalies in Turkey is predominantly carried out on male individuals, while surgical treatment is higher in females than in males.

Current study is a very important resource for retrospectively evaluating 10-year cytogenetic analyses and creating a regional cytogenetic data library in terms of sex development disorders and their causes.

The 5212 cases we scanned include the largest group when compared to similar studies on gender development disorders so far. In addition, the sex-related chromosomal anomalies we detected are consistent with the literature data we scanned in most patient groups. In our study, 154 (92.2%) of the cases who applied with the prediagnosis of azoospermia in men were diagnosed with Klinefelter syndrome.

Table 4: Diagnosis-prediagnosis table of men in terms of gender anomalies.

Prediagnosis (female) N (%)	Normal	Turner S.	Süper F.	Swayer S.	46,X,i(Xq)	46,XX,del(X)	46,tder(X)t(X;21)(q28:q21)mat	45,X/46,Xr(X)	Total
Habitual abortus	855 (59.1)	217 (15.0)	375 (25.9)	-	-	-	-	-	1447
Infertility	229 (75.6)	-	-	-	32 (10.7)	37 (12.4)	-	-	298
Down syndrome	65 (75.6)	-	11 (12.8)	-	-	-	10 (11.6)	-	86
Genetic syndrome	71 (72.4)	14 (14.3)	13 (13.3)	-	-	-	-	-	98
Amenorrhea	116 (93.5)	-	-	8 (6.5)	-	-	-	-	124
Hypergonadotropic hypogonadism	8 (72.7)	3 (27.3)	-	-	-	-	-	-	11
Developmental delay	30 (50)	-	16 (26.7)	-	-	-	-	-	60
Turner syndrome	12 (36.4)	13 (39.4)	8 (24.2)	-	-	-	-	-	33
IVF failure	111 (100)	-	-	-	-	-	-	-	111
Other	173 (74.6)	59 (25.4)	-	-	-	-	-	-	232
Swayer syndrome	1 (100)	-	-	-	-	-	-	-	1
Child with anomalies history	48 (87.3)	7 (12.7)	-	-	-	-	-	-	55
Neuromotor retardation	29 (100)	-	-	-	-	-	-	-	29
Ambiguous genitalia	7 (77.8)	2 (2.2)	-	-	-	-	-	-	9
MR	84 (100)	-	-	-	-	-	-	-	84
Total	1839 (68.7)	315 (11.8)	423 (15.8)	8 (0.3)	32 (1.2)	37 (1.4)	10 (0.4)	14 (0.5)	2678

Table 5: Diagnosis-prediagnosis table of men in terms of gender anomalies.

Prediagnosis/diagnosis N (%)	Normal	Klinefelter syndrome	MixGD	S. Male	46,X,del(Y)(q11)/45,X	46,XY,Yqh+	Total
Habitual abortion	1091 (91.5)	102 (8.5)	-	-	-	-	1193
Azoospermia	13 (7.8)	154 (92.2)	-	-	-	-	167
infertility	190 (54.3)	120 (34.3)	26 (7.4)	-	14 (4.0)	-	350
Down syndrome	94 (100)	-	-	-	-	-	94
Genetic syndrome	102 (90.3)	-	-	11 (9.7)	-	-	113
HH	12 (100)	-	-	-	-	-	12
Developmental ret.	52 (100)	-	-	-	-	-	52
Turner	5 (100)	-	-	-	-	-	5
IVF failure	84 (100)	-	-	-	-	19 (18.4)	84
Klinefelter S.	4 (44.4)	3 (33.3)	-	2 (22.2)	-	-	9
Other	154 (72.3)	59 (27.7)	-	-	-	-	213
Anomaly with child	49 (100)	-	-	-	-	-	49
Neuromotor ret.	23 (100)	-	-	-	-	-	23
Ambiqus G.	13 (100)	-	-	-	-	-	13
MRI	68 (49.3)	70 (50.7)	-	-	-	-	138
Total	1954 (77.1)	508 (20.0)	26 (1.0)	13 (0.5)	14 (0.6)	19 (0.7)	2534

CONCLUSION

Cytogenetic analysis requirement; early diagnosis and treatment in patients with infertility and gender development disorders are important in terms of increasing the life standards of these patients. For this reason, our cases should be directed to chromosome analysis in the early period and followed up with appropriate genetic counselling.

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REFERENCES

1. Jouyan N, Davoudi Dehaghani E, Senemar S, Shojaee A, Mozdarani H. Sex chromosome aneuploidy in cytogenetic findings of referral patients from south of Iran. Iran J Reprod Med. 2012;10(2):141-8.
2. Berglund A, Stochholm K, Claus, Gravholt H. The epidemiology of sex chromosome abnormalities. Am J Med Genet. 2020;184C:202-15.
3. Hall H, Hunt P, Hassold T. Meiosis and sex chromosome aneuploidy: how meiotic errors cause aneuploidy; how aneuploidy causes meiotic errors. Curr Opinion Genet Develop. 2006;16(3):323-9.
4. Rau S, Whitman ET, Schauder K, Gogate N, Lee NR, Kenworthy L, Raznahan A. Behavioral features in Prader-Willi syndrome (PWS): consensus paper from the International PWS Clinical Trial Consortium. J Neurodevelop Disord. 2021;13:61.
5. Thomas NS, Hassold TJ. Aberrant recombination and the origin of Klinefelter syndrome. Human R. 2003;9:309-17.
6. Rafique M, AlObaid S, Al-Jaroudi D. 47, XXX syndrome with infertility, premature ovarian insufficiency, and streak ovaries. Clin Case Rep. 2019;7(6):1238-41.
7. Robinson DO, Jacobs PA. The origin of the extra Y chromosome in males with a 47,XYY karyotype. Human Mol Genet. 1999;8(12):2205-9.
8. Matsuzaki J, Bloy L, Blaskey L, Miller J, Kuschner ES, Ku M, et al. Abnormal auditory mismatch fields in children and adolescents with 47,xyy syndrome keywords xyy syndrome · autism spectrum disorder · magnetoencephalography · vowel mismatch fields · language impairment. Dev Neurosci. 2019;41:123-31.
9. Jacobs P, Dalton P, James R, Mosse K, Power M, Robinson D, Skuse D. Turner syndrome: a cytogenetic and molecular study. Annals Human Genet. 1997;61(6):471-83.
10. Lu X, Wang C, Sun Y, Tang J, Tong K, Zhu J. Noninvasive prenatal testing for assessing foetal sex chromosome aneuploidy: a retrospective study of 45,773 cases. BioMed Central, 2021;14(1):45-9.
11. Tobias ES, Connor JM, Ferguson-Smith M. Essential Medical Genetics. 6th ed. USA: Elsevier; 2011.
12. Al-Alawi I, Goud TM, Al-Harasi S, Rajab A. Cytogenetic studies of 1232 patients with different sexual development abnormalities from the Sultanate of Oman. Reprod BioMed. 2016;32(2):162-9.
13. Balkan M, Akbas H, Isi H, Oral D, Turkyilmaz A, Kalkanli S, et al. Cytogenetic analysis of 4216 patients referred for suspected chromosomal abnormalities in Southeast Turkey. Genet Mol Res. 2010;9(2):1094-103.
14. Duarte AC, Cunha E, Roth JM, Ferreira FLS, Garcias GL, Martino-Roth MG. Cytogenetics of genetic counseling patients in Pelotas, Rio Grande do Sul, Brazil. Genet Mol Res. 2004;3(3):303-8.
15. Singh DN. Cytogenetic study of ındividuals suspected of chromosome anomalies. Clin Pediatr. 1977;16(7): 619-22.
16. Santos CB, Boy RT, Santos JM, Silva MPS, Pimentel MMG. Chromosomal investigations in patients with mental retardation and/or congenital malformations. Genet Mol Biol. 2000;23(4):703-7.
17. Thillainathan S, Sirisena ND, Kariyawasam KWJC, Jayasekara RW, Dissanayake VHW. Cytogenetic analysis of chromosomal abnormalities in Sri Lankan children. World J Pediatr. 2015;11(4):374-9.
18. Froukje MES, Stenvert LSD, Jan CM, Sabine MPF. Long-Term psychological evaluation of ıntersex children. Arch Sexual Behav. 1998;27(2):23-8.

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