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Renal and urinary tract abnormalities associated with chromosome aberrations

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Abstract

The frequency of malformations of the kidney and urinary tract is much higher in patients with chromosome aberrations than in the general population.

Sixty to 100% of “cat-eye” syndrome, 60 to 80 % of Turner, 75% of trisomy 8, 33 to 70 % of trisomy 18, 50 to 60 % of trisomy 13, and over 50 % of triploidy and tetraploidy patients may have such abnormalities.

Renal and urinary tract malformations should be looked for in all patients with chromosome aberrations. Moreover, a chromosome study is indicated in any fetus with an ultrasonographic evidence of urinary tract abnormality. (Int J Pediatr Nephrol 1987; 8: 215–226)

Keywords

Chromosome aberrations; Renal malformations; Urinary tract abnormalities

INTRODUCTION

The association of chromosomal aberrations with multiple congenital anomalies is well known (1,2). The purpose of this paper is to review kidney and urinary tract abnormalities occuring in combination with different chromosome aberrations.

MATERIAL AND METHODS

A thorough review of the genetic and renal literature was performed and our patient files reviewed. Associations of renal and urinary tract abnormalities with chromosome aberrations were classified as autosomal trisomies, autosomal monosomies, sex chromosome aberrations, arid other chromosome aberrations. The chromosome syndromes most commonly associated with renal and urinary tract abnormalities were discussed briefly, and other chromosome conditions with renal involvement were presented in a table form.

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RESULTS

Autosomal Trisomies

1. Trisomy 8 (Warkany syndrome) is characterized by mental retardation, relatively specific facies, absent or dysplastic patellas, joint contractures, plantar/palmar furrows, distinctly abnormal toe posture, vertebral anomalies, narrow pelvis, and ureteral-renal anomalies. It has been proposed that the distal long arm (8q24→8qter) is the critical area that produces the features of trisomy 8 syndrome (3). Renal and urinary tract abnormalities occur in about 75% of cases (3) and include hydronephrosis, bifid pelvis, non-functioning kidney, horseshoe kidney and vesi-coureteral reflex (4–8). These abnormalities occur in 88% of trisomy 8 mosaic syndrome patients (9), and to a much lesser extent in patients with partial trisomy 8 (10).

2. Trisomy 13 (Patau syndrome), which is characterized by cleft lip/palate, polydactyly, micro-phthalmia, CNS and visceral abnormalities, severe mental retardation, and early death, occurs in 1/5,000 to 1/14,500 births (1, 11). Renal abnormalities are present in about 50 to 60% of affected individuals (12, 13). Cystic kidney disease is the most common renal anomaly occurring in over one third of cases, hydronephrosis and hydroureter in about 20%, duplication of the kidney and urinary tract in 7 to 18%, and fetal lobulation of the kidneys in 10% (12, 14). Megacystis, bladder neck stenosis, vesicoureteric obstruction, horseshoe kidney and unilateral renal agenesis are much rarer findings.

3. Trisomy 18 (Edward syndrome), which is characterized by low birth weight, low-set malformed ears, micrognathia, prominent occiput, CNS and visceral abnormalities, rocker bottom feet, overlapping digits, and early death, occurs in 1/4,500 to 1/8,000 live births (1, 15). Renal anomalies occur in 33 to 70% of cases (12, 16) and include duplication of the urinary tract in about 25% of patients, horseshoe kidney in 17 to 24%, cystic kidneys in 8 to 17%, hydronephrosis and hydroureter in 15%, ectopic kidney in 10%, and renal agenesis or hypoplasia in 7%. Rarer abnormalities include unilateral renal hyperplasia, renal hamartoma, fetal lobulation, rotational anomalies, renal dysplasia, and glome-rulosclerosis (12, 13, 16, 17). The association of Wilms ‘ tumor with this syndrome has also been described (18).

4. Trisomy 21 (Down’s syndrome) is the most common autosomal trisomy with an overall incidence of about 1/700 births (1). The association of renal abnormalities with this syndrome is much lower than with trisomy 13 and 18. Four of 141 patiens with trisomy 21 studied by Berg et al (19) had renal abnormalities consisting of renal agenesis or hypoplasia, horseshoe kidney, hypoplastic bladder, and urethral valves. Egli and Stalder (12) found 6.7% of 103 autopsied children with Down’s syndrome to have renal anomalies consisting of renal cysts, hydronephrosis and hydroureter, megacystis, and ureteral stenosis (12, 20). Immature glomeruli and uretero-pelvic junction stricture were also described (20).

Renal and urinary tract abnormalities associated with other trisomic chromosomal conditions are presented in Table 1.

Autosomal Monosomies

1. Chromosome 4 short arm deletion (4p-or Wolf-Wolf-Hirshhorn syndrome). Renal abnormalities occur in about 1/3 of affected patients (32) and consist of renal ageneis or hypoplasia, non-functioning kidney, dilated collecting system, vesicoureteral reflux, and chronic pyelonephritis (12, 64, 65). Hypospadia is a constant finding in this syndrome (32, 64, 66).

2. Chromosome 5 short arm deletion (5p-or Cridu-chat syndrome). The incidence is about 1/50,000 births (1). Ectasia of the distal tubules, horseshoe kidney, duplication of the urinary tract and renal agenesis may occur in about 40% of affected patients, although renal studies in this disorder are few (12, 29).

3. Chromosome 18 long arm deletion (18q-). This disorder is estimated to occur in about 1/60,000 births (67). About 40% of patients have a renal abnormality consisting of polycystic, ectopic or horseshoe kidney, unilateral renal agenesis, hydronephrosis, and hydroureter (12, 59, 68, 69).

   Renal and urinary tract abnormalities as-sociated with other monosomies are presented in Table 2.

Sex Chromosome Aberrations

1. Turner’s syndrome. First described by Turner in 1938 (1, 98), this syndrome is characterized by short stature, ovarian dysgenesis, and sexual infantilism. Incidence is about 1/2,500 newborn females; however, only 3% of the 45,X zygotes survive to term (99). Chromosome findings consist of 45,X or mosaicism for one X and XX, XXX, or XY cell populations.

   Renal and urinary tract abnormalities occur in 60 to 80% of patients with Turner’s syndrome (12, 100, 101); hence, routine investigation of the urinary tract is warranted in all patients with this syndrome. Horseshoe kidney is by far the most commonly reported anomaly occurring in 20 to 45% of patients (12, 101, 102) as compared with 11/400 to 1/750 in the normal population (103, 104). Duplication of the urinay tract occurs in about 20 to 30% of patients, and a rotational anomaly in about 14% (12, 101, 102). Ureteropelvic obstruction; hydronephrotic, ectopic, ptotic, aplastic, hypoplastic or cystic kidney; and urethral meatal stenosis may also be present, but to a lesser extent. Idiopathic hypertension (105) and a double renal artery (106) may also occur. Renal malformations, as well as con-genital abnormalities of the other organs, occur more rarely in mosaic patients than in the classic patient with a 45, X karyotype; however, the type of renal anomaly is similar in both forms of the disease. Over the past two years we have diagnosed 12 cases of Turner’s syndrome, four of whom were mosaics. Two out of these four had renal and urinary tract malformations.

2. Klinefelter syndrome. First described by Klinefelter in 1942 (107), this syndrome is characterized by tall stature, hypogenitalism, hypogonadism, dull intelligence and/or behavioral problems. The incidence is 1/500 newborn males. The karyotype is XXY in 80 % of cases with mosaicism or an extra aneuploidy in the remaining 20% of cases (108). The incidence of urinary tract abnormalities in this syndrome is similar to that in the general population. Renal cysts, hydronephrosis, hydroureter, ureterocele, and chronic glomerulonephritis have been de-scribed in affected patients (12, 109, 110).

Other Chromosome Aberrations

1. Triploidy (69 chromosomes) has an incidence of 1/20,000 births (111), but occurs in 1% of conceptuses (29). It is characterized by growth deficiency, syndactyly of the third and fourth fingers, CNS and visceral abnormalities, a large placenta with hydatidiform changes, and early death. The syndrome usually results from fertilization of a haploid ovum by two spermatozoa. Cystic renal dysplasia and hydronephrosis occur in over 50 % of affected patients (29). Fetal lobulations, pelvic kidney (112), and cystic de-generation of the kidneys (108) have been de-scribed.

2. Tetraploidy (92 chromosomes) is frequently de-scribed in human abortuses, but rarely in living newborns (112, 113). Renal and urinary tract abnormalities may occur in about 50% of cases, and include renal hypoplasia and dysplasia, pyelonephritis, megaureter, vesicoureteral reflex and urethral stenosis (29, 113, 114).

3. “Cat-eye” syndrome is characterized by coloboma of the iris, anal atresia, preauricular tags, and an extra small marker chromosome which is generally thought to be derived from chromosome 22 (duplication 22pter→q11) (115). Renal abnormalities occur in 60 to 100% of these patients and consist of renal agenesis, hypoplasia, and cystic dysplasia; horseshoe and pelvic kidney; ureteropelvic junction obstruction, vesica-ureteral and urethral stenosis, ectopic uretero-vesical orifice, bladder neck obstruction with reflux, abnormal shape of renal pelvis, hypoplastic urinary baldder, and chronic pyelonephritis, (12, 115, 116, 117).

   Renal and urinary tract abnormalities associated with other chromosome aberrations are presented in Table 3.

DISCUSSION

The frequency of fetal malformations is about 2%, with urinary tract abnormalities representing 50% of the total ultrasonographically diagnosed malformations (130). Fifteen percent of these abnormalities are considered to be due to chromo-some aberrations (131). These malformations are due to impaired development of the ureteric bud and metanephrogenic tissue before the 28th day of embryonic development (132). They may result from environmental factors such as maternal illness and exposure to toxic substances, genetic factors or multigenic interactions (133).

The incidence of renal and urinary tract mal-formations is much higher in many forms of chromosome aberrations than in the general population, e.g., 60 to 100% in “cat-eye”, 60 to 80% in Turner, 75% in trisomy 8, 33 to 70% in trisomy 18, 50 to 60% in trisomy 13, and over 50 % in triploidy and tetraploidy syndromes. They occur in 33 to 40% of 4p-, 5p-, and 18q- syndromes, and are much less common in the other chromosome aberration syndromes. The actual incidence is probably higher than what is reported in the literature since many reports do not include investigation of the urinary tract. The available studies show that there is no direct correlation between urinary tract malformations and concomitant mal-formations of other organs, or any particular chromosome aberration (12).

A specific syndrome is usually identified in about 24% of patients with multiple malformations (134). Since 27 to 50 % of these are due to chromo-some aberrations (134, 135) and since 50% of fetal malformations may be due to urinary tract abnormalities (130), a karyotype is indicated in any fetus with ultrasonographic evidence of urinary tract abnormality. Nicolaides et al (136) found chromosome abnormalities in 9 of 39 fetuses (23 %) with ultrasonographic diagnosis of obstructive uropathy. Recently, two fetuses with bilateral renal agenesis and infantile polycystic kidney disease diagnosed by ultrasonography, and studied by the Authors, were found to have 4p- and trisomy 13 syndromes, respectively.

A renal or urinary tract abnormality should be looked for in any patient with multiple congenital anomalies, particularly chromosome aberrations, and a chromosome study should be performed on any fetus with ultrasonographic evidence of urinary tract malformations.

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TABLE 1-

OTHER TRISOMIC CHROMOSOME CONDITIONS ASSOCIATED WITH RENAL AND URINARY TRACT ABNORMALITIES

Chromosome aberrations	Renal abnormality	Reference
1q23 or 5→qter trisomy	Renal cysts and calcification, hydronephrosis	21, 22, 23
2q21→qter trisomy	Ectopic, dysplastic, and horseshoe kidneys; ureteral atresia	24
2q3 trisomy	Vesicoureteral reflux	25
3q2 trisomy	Cystic dysplasia, calcification, and duplication of kidney; accessory kidney	26, 27
4p14→pter trisomy	Renal agenesis, rotated kidneys, intrarenal pelvis	28, 29.
4q2 or 3 trisomy	Cystic, hypoplastic, aplastic, and horseshoe kidney; vesicoureteral reflux; hydronephrosis; hydro ureter	1, 30, 31 32
6p21→pter trisomy	Horseshoe and small kidney, triple renal artery, double renal vein	33
6p2 trisomy	Proteinuria, small kidneys	1, 34
6q21→qter trisomy	Renal dysplasia	35
7q3 trisomy	Hydronephrosis, hydroureter	36
8p trisomy	Ureteral stenosis, absent bladder	1
8q2 trisomy	Hydronephrosis, hydroureter	1
9 trisomy	Cystic kidneys	37
9 trisomy mosaicism	Renal cysts	38
9p trisomy	Hydronephrosis, horseshoe kidney	39
9q3 trisomy	Hydroureter	1
10 trisomy mosaicism	Immature glomeruli	40
10p11→pter trisomy	Rotated, aplastic, cystic, dysplastic, and double kidneys; megaureter; aplastic ureter	41,42,43 44, 45
10q2 trisomy	Renal failure; hypoplastic, dysplastic, cystic, hydronephrotic and double kidneys; double collection system	46, 47 48, 49
11 p15 trisomy	Large kidneys, Wilms'tumor	50
11q2 trisomy	Renal agenesis, vesicoureteral reflux	51, 52
12q2 trisomy	Hydronephrosis, ureterocele, pelvic kidney	53, 54
12q24.1→qter trisomy	Ureterocele, hydronephrosis, ectopic and aplastic kidney	53,54 55
13q2 or 3 trisomy	Double renal artery, unspecified renal abnormalities	56, 57
14 trisomy mosaicism	Renal failure	1
17q21→qter trisomy	Hypoplastic and cystic kidneys, hyperplastic urinary bladder	58

Chromosome aberrations	Renal abnormality	Reference
17q23→qter trisomy	Unspecified urinary tract malformations	1
18pter→18q21.2 trisomy	Multicystic kidneys, ureteral agenesis	59
18q2 trisomy	Polycystic and ectopic kidneys, vesicoureteral reflux, hydroureter, unspecified renal malformations	1, 59, 60
19q13→pter trisomy	Hydronephrosis; hydroureter; ectopic, malrotated or cystic kidneys	61, 62
20p trisomy	Hydronephrosis, polycystic kidneys, double collecting system	63

TABLE 2-

OTHER MONOSOMIC CHROMOSOME CONDITIONS ASSOCIATED WITH RENAL AND URINARY TRACT ABNORMALITIES

Chromosome aberrations	Renal abnormality	Reference
1q42 or 43→qter monosomy	Solitary kidney, vesicoureteral reflux	70
3p11→p14.2 monosomy	Horseshoe kidney	71
4q31→qter monosomy	Ectopic kidney, double collecting system	72, 73
4q3 monosomy	Double collecting system, hydronephrosis, lobulated kidneys	72, 74, 75
5q13→q22 monosomy	Horseshoe kidney	76
6q13→q15 monosomy	Ectopic kidney	77
7q13-p21 monosomy	Hydronephrosis, renal dysplasia, ureteral diverticuli, ureterocele, double collecting system	78, 79 80
10p13→pter monosomy	Cystic and segmental renal dysplasia, double collecting system, hydronephrosis, hydroureter	81,82 83
11p11 monosomy	Horseshoe kidney	84
11p13 monosomy	Wilms' tumor, malrotated kidney, pyelonephritis, hypertension	85, 86, 87
11q22 or 23→qter monosomy	Renal duplication, hydronephrosis	88
11q2 monosomy	Multicystic kidneys, double collecting system, hydronephrosis	89, 90
13q monosomy	Vesicoureteral obstruction, hydronephrosis, hydroureter	12
13q3 monosomy	Hypoplastic kidneys	1
15q22→q24 monosomy	Cystic renal dysplasia	91
17p11.2 monosomy	22% of patients may have enlarged or solitary kidney, hydroureter, hydropelvis, or malpositioned vesicoureteral junction	92
18p monosomy	Unspecified renal malformations	67
21q monosomy	Aplasia, dysplasia, and abnormal shape of kidney	93, 94, 95
22q12 monosomy	Dysplastic and cystic kidneys	96, 97

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TABLE 3-

OTHER CHROMOSOMAL ABERRATIONS ASSOCIATED WITH RENAL AND URINARY TRACT ABNORMALITIES

Chromosome aberrations	Renal abnormality	Reference
2q3 trisomy/7p22 monosomy	Renal hypoplasia	118
9p tetrasomy	Hypoplastic kidney	119
18p tetrasomy	Mai rotated or horseshoe kidney, double ureter	120,121 122
r(10)	Renal failure, hydronephrosis, hydroureter	123, 124
r(13)	Aplastic, hypoplastic or ectopic kidney	12, 125 126
r(15)	Incomplete duplication of kidney	127
r(18)	Ectasia of proximal tubules, megaureter, hydronephrosis, vesicoureteral obstruction	12, 52
r(21)	Renal agenesis, ureteral anomaly	1
Small marker chromosome of unknown origin	Hypoplastic kidney, hydronephrosis, vesicoureteral reflux	128
49, XXXXX	Renal hypoplasia and dysplasia	129