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Congenital Malformations in Chromosomal Abnormalities: Analysis of 18 Autopsy Cases

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=Abstract=To determine the structural anomalies associated in various chromosomal aberration syndromes we have analyzed postmorm findings of 18 autopsies with chromosomal anomalies that were collected during the last 14 years, 1975-1988. The diagnosis was confirmed by karyotyping in 14 cases and characteristic sets of anomalies in 4 cases.

The cases consisted of 10 cases of Edwards syndrome, 5 cases of Down syndrome, 2 cases of Patau syndrome and one case of 4p-syndrome. Four cases were stillborns and 14 cases survived from a few hours to months. There were 3 males and 15 females. Cardiovascular malformations were common. They were patent ductus arteriosus, atrial septal defect and ventricular septal defect. They were particularly common in Down and Edwards syndrome. Among respiratory anomalies, abnormal lobation was common. Tracheo-esophageal fistula and lung hypoplasia were also seen in some cases. They were particularly common in Edwards syndrome. Gastrointestinal malformations were also frequently encountered in Edwards syndrome. They were diaphragmatic hernia, Meckel's diverticulum and mobile intestine. Genitoruinary malformation consisted of cystic dysplasia, horseshoe kidney and hypoplasia of gonads. These anomalies were also common in Edwards syndrome. Central nervous system anomalies were common in Patau syndrome. Holotelencephaly and microcephaly were common. Other skeletal and characteristic facial anomalies were also seen in these malformation sydrome.

It was concluded that a fairly characteristic constellation of structural defects were associated with specific type of chromosomal abnormalities. Therefore these sets of findings could be applied as diagnostic criteria when chromosomal analysis is not available or fails.

Key words: ***Chromosome, Malformation, Trisomy syndrome, Edwards syndrome, Down syndrome, Patau syndrome***

INTRODUCTION

It has been known since 1959 that certain chromsomal aberrations are associated with syndromes of congenital malformations(Patau 1960; Edwards 1960; Finley *et al.* 1963; Butler *et al.* 1963; Bergsma 1979). In korea there seems to be comprable number of patients with chromosomal disorders as compared to other countries although they consisted mainly of sporadic case reports(Kang *et al.* 1986). For the malformation of the internal organs there were few careful studies except for analyses of congenital heart disease in patients with Down syndrome(Park *et al.* 1986).

To determine the structural anomalies associated in various chromosomal aberration syndromes we have analysed autopsy findings on a total of 18 cases of chromosomal abnormalities

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and listed the various type of malformations according to organ system and compared with the data described in the literature.

MATERIALS AND METHODS

A total of 18 cases of chromosomal disorders were collected from the autopsy files of the Department of Pathology, College of Medicine, Seoul National University and Seoul National University Children's Hospital between 1975 and 1988. They consisted of 10 cases of Edwards syndrome, 5 cases of Down syndrome, 2 cases of Patau syndrome and a case of 4p-syndrome. One case of Patau syndrome(case 16) was previously reported by one of us(Chi *et al.* 1979). Cytogenetic studies of 16 cases were performed on lymphocyte cultures from the blood of patients while alive or at the time of autopsy. There were a total of 14 sucessful analyses. Chromosomal study failed in cases 16 and 5 because of the prolonged interval of intrauterine death. Though no chromsomal studies were done in case 11 and 10, the anomalies present in these cases were compatible with those of trisomy 18 and 21 syndrome. The results of chromosomal studies are presented in Table 1. We have analysed the external and internal anomalies of all cases by reviewing autopsy records of the patients and photographs which had been taken at the time of autopsy. The pathologic findings on all 18 cases are presented in tabular form.

RESULTS

1. Clinical features

The clinical findings inlcuding patient's age, sex, gestational age, birth weights, maternal age, drug medication and pregnancy complication are summarized in Table 1. The survival period was generally short. Two patients with Patau syndrome were born dead. The age of patients with Edwards syndrome ranged from 16 hours to 21 days with a mean of 4 days. The patients with Down syndrome were relatively long lived and one of them died of leukemia at 4 months of age. Female to male ratio was 15:3 with a preponderance in the female. Except for two patients with Edwards syndrome, all were products of fullterm gestation, and most of them were small for their gestational ages. Maternal ages of the patients ranged from 22 to 36 years with an average of 28 years. The mean maternal age was higher in Edwands syndrome group, 29.5 years as compared with 28 years for Down syndrome and 27 years for Patau syndrome. The pregnancy of five cases were complicated by polyhydramnios.

2. Pathologic characteristics

1) Malformations of the cardiovascular system

A great variety of malformations was encountered and are listed in Table 2. In all patients except case 11 cardiovascular anomalies were found. Twelve patients had combination of anomalies. Ventricular and atrial septal defects and patent ductus arteriosus were the most common malformations in Down and Edwards syndromes. Patent ductus arteriosus was the most common anomaly overall, and always combined with other cardiac defects. Single umbilical artery and bicuspid pulmonic valve were relatively frequent lesions. In five hearts only a single lesion was noted, representing patent ductus arteriosus, ventricular septal defect, left superior vena cava and single umbilical artery. The following lesions were observed only once: interrupted aortic arch, left superior vena cava, tetralogyof Fallot, abnormal origin of carotid artery, mesocardia and dextrocardia.

2) Malformation of the respiratory tract

Malformations of the respiratory tract are listed in Table 3. The most common anomalies were abnormal lobation of the lungs, consisting of two or four lobes of the right lung. Two patients with Edwards syndrome had tracheoesophageal fistula with esophageal atresia (Type C). There was hypopasia of the lung in 3 cases of Edwards syndrome. A case of choanal atresia was seen in Edwards syndrome.

3) Malformtion of the Gastrointestinal Tract

Malformations listed in Table 4 occurred in 8 cases of Edwards syndrome, and in one each of Down and Patau syndrome. Of 8 patients with Edwards syndrome 5 showed combined anomalies of the alimentary tract. Disphargmatic defect, Meckel's diverticulum and mobile large intestine were the leading malformations. The following lesions were found only once; hypoplastic gallbladder, abonormal lobation of the liver and spleen, accessory spleen, agenesis of the anus and mesoliver. Cystic dilatation of the pan-

Table 1. Clinical summary of 18 autopsy cases of chromosomal disorders

Case No (Autopsy No)	Age/Sex	Gestational age (wks)	Birth weight (kg)	Karyotype	Maternal age (yrs)	Obstetric history	Remark
1 (A83-29)	2d/M	38	2.2	47, XX, +18/46, XY	36	aspirin	
2 (A84-42)	13d/F	43	1.7	47, XX, +18	25		
3 (A84-56)	3d/F	43	2.5	47, XX, +18	28	polyhydramnios	generalized seizure
4 (A85-16)	1d/M	35	1.9	47, XY, +18	32		
5 (A85-24)	16h/F	44	3.0	failed	27	polyhydramnios	congenital pneumonia
6 (A85-28)	20h/F	40	1.7	47, XX, +18	30	polyhydramnios	meconium asp. pneumonia
7 (CHA86-13)	1d/F	32	1.2	47, XX, +18	26		
8 (CHA86-21)	1d/F	41	1.9	47, XX, +18	27	polyhydramnios	
9 (CHA86-45)	SB/F	39	3.4	47, XX, +18	23		
10 (CHA87-107)	21d/F			note done		a	
11 (A82-107)	4d/F	39	3.8	note done	31		
12 (CHA86-34)	4M/F	40	unknown	47, XX, +21			cogenital megakaryoblastic leukemia
13 (CHA87-25)	13d/M	39	3.1	47, XY, +21	30		congenital megakaryoblastic leukemia
14 (CHA87-39)	T/F	23	1.2	47, XX, +21	30	amniocentesis	
15 (CHA88-28)	13d/F	39	2.5	47, XX, +21	27		transient myeloproliferative disorder
16 (RCM #31)*	SB/F	41	1.95	failed	26		
17 (CHA87-36)	SB/F	41	1.4	47, XX, +13	28	polyhydramnios	hydorcephalus & fetal ascites on US
18 (OA85-18)	2M/F	40	1.8	46, XX, 4-	22	breech	pneumonia

*Published cases: RCM, registry of congenital malformation; SB, Stillbirth; T, Termination; US, Ultrasonography

Table 2. Cardiovascular malformations in chromosomal disorders (18 autopsy cases)

Diseases	Edwards syndrome (n=10)	Down syndrome (n=5)	Patau syndrome (n=2)	4p syndrome (n=1)	Total (n=18)
Patent ductus arteriosus	6	2	2		10
Ventricular septal defect	5	1			6
Atrial septal defect	3	3		1	7
Patent formamen ovale	2				2
Bicuspid pulmonic valve	2				2
Tetralogy of Fallot			1		1
Interrupted aortic arch	1				1
Abnormal carotid artery			1		1
Left superior vena cava	1				1
Dextrocardia			1		1
Mesocardia				1	1
Globular heart			2		2
Single umbilical artery	3				3

Note: n; number of cases

Table 3. Anomalies of respiratory system in chromosomal disorders

Diseases	Edwards syndrome (n=10)	Down syndrome (n=5)	Patau syndrome (n=2)	4p⁻ syndrome (n=1)	Total (n=18)
Tracheo-esophageal fistula with esophageal atresia	2				2
Abnormal lobation of lung	5	1	1		7
Choanal atresia	1				1

Table 4. Gastrointestinal malformations

Diseases	Edwards syndrome (n=10)	Down syndrome (n=5)	Patau syndrome (n=2)	4p syndrome (n=1)	Total (n=18)
Diaphragmatic defect	3				3
Meckel's diverticulum	2				2
Mobile intestine	2				2
Agenesis of anus	1				1
Abnormal lobation of liver	1				1
Mesoliver	1				1
Hypoplasia of gallbladder	1				1
Cystic change of pancreas	1				1
Abnormal lobation of spleen	1				1
Accessory spleen	1	1			2
Heterotopic splenic pancereas			1		1
Meconium peritonitis	1		1		2

Table 5. Malformations of genitourinary system

Diseases	Edwards syndrome (n=10)	Down syndrome (n=5)	Patau syndrome (n=2)	4p⁻ syndrome (n=1)	Total (n=18)
Horseshoe kidney	2		1		3
Cystic kidneys	7	1			8
Albsence of fetal lobulation of kidney				1	1
Double collecting system	1				1
Hydroureter	1				1
Bladder diverticulum	1				1
Rectovesical fistula	1				1
Ambiguous genitalia	2				2
Cryptorchidism	1				1
Hypoplasia of ovary	3				3
Hypoplasia of uterus	1				1
Bicornuate uterus	1				1
Abnormal adnexa			1		1

creatic duct and nests of respiratory epithelia in the throid were noted in one each with Edwards syndrome. In the spleen of one patient with Patau syndrome heterotopic pancreatic tissue was observed. Meconium peritonitis occurred in one case of Edwards and Patau syndrome each.

4) Malformation of the urogenital system

Urogenital anomalies found in this series are listed in Table 5. The leading anomalies were cystic kidneys, including microcysts and horseshoe kidneys. There were 3 horseshoe kidneys and 8 cystic kidneys. One case of cystic kidneys was associated with hydroureter and bladder diverticulum. Double collecting system of the ureter and pelvis was combined with one case of horseshoe kidney. The kidney of a patient with 4p⁻syndrome had no fetal lobulation. Hypoplasia of the ovaries occurred in 3 cases, bicornuate uterus in two, enlargment of the clitoris in two, and hypoplasia of the uterus, abnormally located adnexa, rectovesical fistula in one case each.

5) Malformation of the central nervous system

Malformations of the central nervous system, as shown in Table 6, were less common in Edwards than in Patau or 4p- syndrome. Nine of 18 cases of chromosomal abnormalities showed anomalies of the central nervous system, and of these 6 had combination of anomalies. The brain was a small and globular with short anteriopos-terior diameter in 6 cases of chromosomal abnormalities. representing 4 cases of Edwards syndrome, and one each of Down and Patau syndrome. Abnormal gyri were noted in cases of Edwards syndrome and included simplification and deficiency of the gyrus. Two (Case 6 & 8) among 5 patients with Edwards syndrome showed multiple anomalies of the central nervous system. Case 8 showed agenesis of the corpus callosum, hypoplasia of the cerebellum and pyramid, abnormal configuration of the hippocampus, simple gyri, and a small globular brain. Case 6 showed a small olivary nucleus and heterotopic granular cells in the cerebellar white matter and heterotopic glial islands in the leptomeninges. Two patients with Patau syndrome showed the spectrum of holotelencephaly. One was of the semilobar type and another of the labor type. The former was associated with deficiencies of the olfactory bulb, lamina cribrosa, crista galli and pituitary. A case of 4p-syndrome showed a small globular brain, abnormal configuration of the hippocampus and an intraventricular cyst.

6) Malformations of the skeletal system

The skeletal malformation listed in Table 7, occurred mainly in the Edwards and Patau syndrome. The well known typical flexion contracture of fingers were by far the most common

Table 6. Malformation of central nervous system

Diseases	Edwards syndrome (n=10)	Down syndrome (n=5)	Patau syndrome (n=2)	4p⁻ syndrome (n=1)	Total (n=18)
Box-shaped brain	4	1		1	6
Abnormal gyri	2				2
Microcephaly			2		2
Holotelencephaly			2		2
Heterotopic neuroglial cells	2				2
Hypoplasia of cerebellum	1				1
Hypoplasia of pyramid	1				1
Agenesis of corpus callosum	1				1
Agenesis of olfactory bulb			1		1
Abnormal hippocampus	1			1	2
Abnormal olivary nucleus	1				1
Intraventricular cyst				1	1
Hydrocephalus	1				1

Table 7. Skeletal malformations

Diseases	Edwards syndrome (n=10)	Down syndrome (n=5)	Patau syndrome (n=2)	4p⁻ syndrome (n=1)	Total (n=18)
Flexion deformity					
of fingers	9	2			11
of joint	3				3
Overlapping fingers	6		1		7
Short 5th finger		2			2
Distally implanted thumb	1				1
Rocker bottom feet	9	1	2		12
Hypoplasia of toe	7				7
Hypolasia of nail	8	1	1		10
Narrow pelvis	1				1
Congenital hip dislocation	1				1
Syndactyly			1		1
Polydactyly			1		1
Hypoplasia of muscle	1				1
Interdigital web	1				1
Simian crease	5	2	1	1	9
Radio-ulnar synostosis				1	1
Abnormal ribs & vertebrae				1	1

lesions and occurred in 9 cases of Edwards syndrome and 2 cases of Down syndrome. This typical findings was characterized by flexion deformities of the fingers with abduction of the thumb across the palm and index finger with overriding one or more of the other fingers, and the fifth finger overridig the fouth. The protuberance of the heels, a so-called "rocker bottom feet" was present in 12 cases. Seven patients showed shortness of the dorsiflexed and hypoplastic first toe. Hypoplastic nails were seen in 10 cases. Simian crease occurred in 9 cases. Of

Table 8. Craniofacial anomalies

Diseases	Edwards syndrome (n=10)	Down syndrome (n=5)	Patau syndrome (n=2)	$4p^-$ syndrome (n=1)	Total (n=18)
Ocular malformation					
Hypertelorism	5	1		1	7
Hypotelorism	1		2		3
Absent eyebrow			1		1
Oblique palpebral fissure		5			5
Epicanthc folds		2			2
Microcornia	1				1
Congenital cataract	1				1
Coneal opacity	2				2
Iris colobomata			1		1
Aural malformation					
Low-set ear	10	2	2		14
Malformed ear	10	2	2		14
Nose anomalies					
Flat nasal bridge	1	4		1	6
Beaked nose	5				5
Small nose		3			3
Absent nose			2		2
Absent nasolabial fold		1			1
Cranofacial					
Prominent occiput	9				9
Prominent forehead				1	1
Flat forehead		1			1
Craniolacunia	1				1
Micrognathia	4	1			5
Receding chin	10	1		1	12
Cleft lip & palate	1		2		3
High arched palate	4			1	5

these 9 cases 5 occurred in Edwards, 2 in Down syndrome and one in Patau and 4p-syndrome. Anomalies observed only once were narrow pelvis, congenital hip dislocation, loss of calf muscle mass, interdigital web, radio-ulnar synostosis, distally implanted thumb, pairs of abnormal rib and vertebrae. Two patients with Down syndrome showed a short little finger.

7) Cranio-facial anomalies

The cranio-facial abnormalities are listed in Table 8. The facial features of patients were characteristic for each type of chromosomal abnormality. The face of Edwards syndrome appeared small and triangular with a prominent occiput(90%) and small receding chin(100%). The patients with Down syndrome showed a flat facial profile with oblique palpebral fissure(5 cases) prominent epicanthal folds(2 cases) and flat small nose(4 cases). The facial anomalies of Patau syndrome were characterized by microcephaly, cleft palate and lip, absence of nose and nasal bone, and low-set malformed ears. One case of 4p- syndrome had microcephaly, prominent forehead, hypertelorism, flat small nose, mirognathia, and malformed ears. Anomalies of the eye were relatively common. Hypertelorism was noticed in 5 cases of Edwards syndrome and in a case of Down and 4p-syndrome. Hypotelorism occurred in one case of Edwards and in two patients with Patau syndrome. In Edwards syndrome microcornia and congenital cataract were seen in one case each and corneal

opacity in 2 cases. Brushfield spots occurred in one patient with Down syndrome and colobomata in the iris in one patient with Patau syndrome. The latter had no eyebrows. A great variety and numbers of malformations of the outer and inner ears was noted. Low-set ears were found in all cases of Edwards syndrome and in 2 cases of Down and Patau syndrome each. Malformed ears were noticed in 14 cases of chromosomal abnormalities. Of these 14 cases 10 occurred in Edwards syndrome and 2 in Down and Patau syndrome, each. There was great variability in the shape of the ear from patient to patient and from oen side to the other. The small hypoplastic ears were noticed in 3 of 14 malformed ears. Among cases of Edwards syndrome cleft palate occurred in 1 case, and narrow and high arched palate in 4. A case of high arched palate occurred in the 4p- syndrome. Short neck occurred in 3 cases of Edwards syndrome and in 1 cases of Down syndrome.

8) Blood abnormalities

Among 5 patients with Down syndorme 3 had blood abnormalities. Congenital megakaryoblastic leukemia occurred in 2 cases and transient myeloproliferative diseases in 1 cases. On postmortem examination of 2 patients with leukemia, all visceral organs were affected by the tumor cells and included liver, spleen, bone marrow, heart, lung, kidney, and adrenal and pancreas. The tumor cells consisted of immature megakaryoblasts and abnormal megakaryocytes. One patient with transient myeloproliferative disease displaced no abnormal cells in any organ.

DISCUSSION

We have described the clinicopathologic findings that have been observed by postmortem examination of patients with chromsomal abnormalities. It is demonstrated that chromosomal disorders produce various types of cogenital malformations affecting all the organs system of the body. Association anomalies are more common and wide ranging in Edwards and Patau syndrome than in Down syndrome. The patients with Down syndrome are apparently normal except for a characteristic facial profile with oblique pabebral fissure, prominent epicanthic folds, and small flat nose. Patients with Edwards syndrome show small and triangular faces because of a relatively large head and a small receding chin. The skull regularly shows occipital prominence. These facial features seem to be characteristic of Edwards syndrome. The external ears are low-set, abnormally large and deformed or rudimentary. Flexion coontracture of fingers with overlapping, rocker bottom feet and shortness of the dorsiflexed first toe are more commonly found in patients with Edwards syndrome. A combination of the above anomalies serve to distinguish children with Edwards syndrome from other trisomics.

Although virtually similar anomalies are found in both 13 and 18 trisomics, the constant features, especially external appearance of each are different enough that discrimination between two is not difficult. In patients with Patau syndrome it is characterized by microcephaly, midline cleft lip and palate, hypotelorism, microphthalmia, absence or rudimentary of the nose. These facial anoamlies are well known to be intimately related to anomalies of brain that are induced by the failure of clevage of the developing prosencephalon. The brain anomalies vary from alobar holotelencephaly to lobar holotelencephaly to a normal brain. (Warkany 1971). The central nervous system is severely defective in most cases of Patau syndrome. Warkany *et al.* (1966) reviwed reports of 32 autopsies and showed the most frequent malformations noted below. The holotelencephaly metioned varied greatly in extent. Some patients showed fused frontal lobes and single vetricles suggestive of the cylopia series, whereas in others only deficency of the olfactory nerves and lobes were metioned. Two cases of Patau syndrome reported in the Korean literature showed the cyclopia series (Kim 1987). Besides holotelencephaly cerebellar anomalies, corpus callosum defect and hydrocephaly were also frequently metioned. Malformation of the cardiovascular and urogenital systems are not rare but they were not as conspicuous as defects of the central nervours system. On Warkany's review of Patau syndrome no horsehoe kidney has been reported, but in our series a case of horseshoe kidney was noted. Flexion contracture of the fingers are an outstanding symptom, as in trisomy 18, but polydactyly is caracteristic of trisomy 13. In Warkanys' review of 40 recorded cases, polydactyly occurred in 28.

In the korean literature 1621 cases of chromo-

somal disorders have been reported. Most of them were sporadic cases consisting of descriptions of especially craniofacial and limb anomalies. Kim(1987) reviewed these reported cases of chromosomal abnormalities and analysed the clinical findings according to each type. His review shows that the most common finding noted in patents with Down syndrome is epicanthal folds occurring in 91% of cases. Oblique palpebral fissure, short 5th fingers, high arched palate, brachycephaly, flat nose and face are the leading findings occurring in similar incidence to those of other countries and our series. Brushfield spots though occurring in 35-85% of children with Down syndrome were found in only two cases including one case in our series. Congenital heart disease is an important feature of Down syndromes, but reliable estimates of its frequency are hard to find, since they vary according to the age of patients and with diagnostic method. The most serious cardiovascular anomalies result in early death and are, therefore not encountered in older patient. Even in the autopsy series there are marked variations according to the age distribution. Atrioventricular communis is reported to be typical for Down syndrome by some authors (Rowe & Uchida 1961; Greenwood and Nadas 1976; Park *et al.* 1977), whereas in Warkany's(1966) and Park's analyses(1986) ventricular and atrial septal defects and patent ductus arteriosus are more common than atrioventricular communis. In our series atrial septal defect, patent ductus arteriosus and ventricular septal defect are the most common anomalies. The number of these cases is not sufficient to draw comparison as to frequency of cardiac anomalies in Down syndrome. It is well established that leukemia is 3 to 15 times more common in children with Down syndrome than in the general population. Many cases of this association have been reported in recent years and there are two cases of leukemia occurring in patient with Down syndrome in the Korean literature. One of them is congenital acute myelocytic luekemia in a twelve day old male neonate with Down syndroome (Chi *et al.* 1972). The hematologic picture of leukemia in Down syndrome is variable, thus the type of leukemia is not specific. Acute lymphogeneous or myelogeneous leukemia is the most frequent type of leukemia. There are no available reports on congenital megakaryoblastic leukemia ssociated with Down syndrome, as noted in this report.

There are reported 45 cases of Edwards syndrome in the Korean literature. Anomalies described in more than 50 percent of 45 cases are as follows: Low-set and malformed ears, prominent occiput, high arched palate, web neck, distant nipple, dorsiflexed toes, rocker bottom feet, flexion deformity, limited hip abducton, small pelvis, shield chest, micrognathia and ocular malformation. These anomalies occur in the same incidence as in our series. But the incidence of cardiac anomalies is less than in our own and Warkany's series. Interrupted aortic arch which is not noted in Warkany's series is found in a cases of Edwards syndrome in our series. The incidence of other associated anomalies affecting the genitoruinary and gastrointestinal tracts in our series are comparable with the figures of Warkany's series, but higher than those of Kim's review. This discrepancy of the occurrence between Kim's and our series may not represent the actual difference in this country. It may result from that the majority of cases reported in Korea are based on the external anomalies detected on physical examination of patients. Therefore It seems to be of value to describe malformations of the internal organ revealed on postmortem examination of patients with chromosomal abnormalities. Despite variation in the external appearance consideration of the facial profile together with the other anomalies permit a resonably accurate diagnosis, when chromosomal analysis is not available or fails.

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= 국문초록 =

염색체 이상 증후군의 병리학적 검색

서울대학교 의과대학 병리학교실

서연림 · 지제근

염색체 증후군에 수반되는 각종 구조적 이상을 규명하기 위하여 1975년부터 1988년 사이에 서울대학교병원에서 부검한 18예의 염색체 이상증후군을 토대로 하여 병리학적 검색을 시행하였다. 진단한 14예에서 염색체 분석을 통하여 그리고 나머지 4예에서는 특징적인 동반기형을 통하여 확인되었다.

18예의 염색체 이상증후군은 Edward증후군이 10예, Down증후군이 5예, Patau증후군이 2예 그리고 4p 결손 증후군이 1예였다. 18예중 4예는 사산아였고 14예는 수시간에서 4개월까지 생존하였다. 동반된 심맥관 기형은 동맥관 개존증, 심방 및 심실 중격 결손이 가장 흔했고 특히 Down과 Edwards증후군에서 현저했다. 호흡기 기형은 분엽이상이 흔했고 그외에 기관식도루와 폐저형성증이 있었고 특히 Edwards증후군에서 현저하였다. Edwards증후군에서는 횡격막 결손과 Meckel게실등의 소화계 기형이 흔했다. 비뇨생식기 기형으로는 신장의 낭성 이형성, 마제신 그리고 생식선의 저형성등이 있었다. 중추신경계 기형은 Patau증후군에서 흔했고 전종뇌증과 소두증이 대부분이었다. 기타 골격 및 특징적 안면 기형등이 동반되었다.

이상 소견을 종합하건대 상기 염색체 이상증후군에 있어서는 형태학적으로 특징적 기형의 조합을 이루었고 따라서 이러한 소견들은 핵형 검사를 못하거나 실패하였을때 진단에 도움이 된다고 판단되었다.