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THE PREVALENCE AT BIRTH OF CONGENITAL MALFORMATIONS AT A MATERNITY HOSPITAL IN OSAKA CITY, 1948–1990

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Summary The frequencies of congenital malformations at St. Barnabas' Hospital, Osaka were studied during the period 1948–1990. There were 1,418 malformed infants diagnosed during the first week of life among a total number of 131,996 births during the period. Of the 1.07% of malformed children, 0.90% had single and 0.17% had multiple malformations. The birth prevalence was 1.07% in singletons and 1.20% in twins. There was significant difference in prevalence between males (1.15%) and females (0.97%). The prevalence was 0.88% for the period 1948–1958 and increased with each yaer cohort up to the recent period when the value was 1.38%. Varus deformities of feet was the most common defect (23.4 per 10,000 births), followed by polydactyly (finger) (7.8), anencephaly (7.1), cleft lip and palate (6.5), syndactyly (toe) (6.5), cleft lip (6.3), and Down's syndrome (6.1). The prevalence of conjoined twins was 1 in 65,551 deliveries. Significant differences occurred between males and females for cleft palate, cleft lip and palate, syndactyly (finger), oligodactyly (finger), and hypoplastic auricle. The prevalences for valgus deformities of feet, syndactyly (toe) and atresia ani increased significantly with the year cohort, whereas the prevalence of cleft palate decreased significantly with the year cohort. Overall prevalence was significantly higher in 1970–1990 (1.28 %) than in 1948–1969 (0.88%). Similar results were examined for cleft lip and palate, syndactyly (finger), syndactyly (toe), hypospadias, hydrocele testis, and Down's syndrome.

Key Words congenital malformation

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INTRODUCTION

During the past two decades monitoring systems for congenital malformations have been developed in Japan. Data are collected from hospitals or from population-based systems. Since 1972 the Japan Association for Maternal Welfare (JAMW) has been investigating 76 types of congenital malformations from about 238 hospitals (Sumiyoshi *et al.*, 1989). They determined a prevalence of 0.82% during the period 1972–1987. Kato *et al.* (1990), who have been investigating the prevalence of malformations at Tokyo Metropolitan Hospitals since 1979, determined a prevalence of 1.5% during the period 1979–1988.

The study reported here concerns secular changes in the prevalence of congenital malformations in newborns at a maternity hospital during the period 1948–1990. A preliminary analysis based on a part of the present study was published by Yamamura *et al.* (1977).

MATERIALS AND METHODS

The prevalence of malformations, diagnosed during the first week of life, at the St. Barnabas' Hospital, Osaka City were studied during the period 1948–1990. There were 1,418 malformed children (785 males, 619 females, and 14 with undetermined sex) diagnosed from a total number of 131,996 consecutive births: 129,734 live births and 2,262 fetal deaths. Table 1 presents a summary base data. The sex ratio was 1.07 during the period. The stillbirth rate was 17.1 per 1,000 births. Among 131,102 deliveries, there were 885 multiple deliveries: 877 twin pairs, 7 triplet sets, and 1 quadruplet set.

Data on congenital dislocation of the hip and the specific cardiovascular defects

Table 1. Summary of the base data for the present study, 1948–1990.

All births	131,996 (131,102 deliv.)		
Single	130,217		
Twins	1,754 (877 deliv.)		
Triplets	21 (7 deliv.)		
Quadruplets	4 (1 deliv.)		
Sex	Live births	Stillbirths	Total
Male	67,078	1,168	68,246
Female	62,624	1,038	63,662
Unknown	32	56	88
Total	129,734	2,262	131,996
Sex ratio:	107.2		

were eliminated from the present analysis because of the difficulty in making a definite diagnosis of these at birth.

Each record has the baby's sex, date of birth (live birth or fetal death), maternal age, parity, birth weight, and gestational age.

RESULTS

Secular changes in the prevalence of malformations

A complete listing of the frequency of each malformation is given in the Appendix. The malformations are presented by system (or type) and sex. There were 1,751 malformations, of which 1,193 were single and 558 multiple malformations; the latter was counted as many times as they occurred in an individual. The overall prevalence was 1.33%; 1.46% for males and 1.15% for females. Among all births, 1,418 babies (1.07%) had malformations: 1,192 (0.90%) with single and 226 babies (0.17%) with multiple malformations. The proportion of the infants with multiple malformations among all those malformed was 17% (133/785) for males, 14% (85/619) for females, and 16% overall. The difference between the sexes is not significant.

Table 2 shows the number of malformed cases and the prevalence according to sex and birth year cohorts. The total number of births and malformations were divided into four periods: 1948–1958, 1959–1969, 1970–1980, and 1981–1990. The corresponding proportions of multiple malformed cases among all malformed cases were 0.14, 0.16, 0.18, and 0.15, respectively. The prevalence was 0.88% during the period 1948–1958, and increased with each cohort up to the most recent period (1981–1990) where the value was 1.38%. The regression coefficient of prevalence on the birth year cohort is significant at the 5% level. The overall prevalence is higher in males (1.15%) than in females (0.97%) and this difference is significant.

Table 3 shows the secular changes in the prevalence of selected malformations during the period 1948–1990. The first report of Down's syndrome occurred in 1961, consequently no prevalence data are available for the earliest period 1948–1958. Then the prevalence rapidly increased with the year cohort: 3.8 per 10,000 births for 1961–1969, 5.6 for 1970–1980, and 10.5 for 1981–1990. The prevalence of each of valgus deformities of feet, syndactyly (toe), and atresia ani increased significantly with the year cohort. However the prevalence of cleft palate decreased significantly with the year cohort. The prevalence of each of anencephaly, spina bifida, congenital hydrocephaly, cleftlip, cleft lip and palate, varus deformities of feet, polydactyly (finger), polydactyly (toe), syndactyly (finger), hydrocele of the testis, and hypospadias remained constant with the year cohort.

The prevalence of selected malformations

Table 4 shows the numbers and of prevalence by sex of selected malformations during the period 1948–1990. To compute the prevalence of Down's syndrome,

Table 2. Numbers and prevalence of congenital malformed cases by sex and by birth year cohorts, 1948–1990.

Period	Number of malformed cases				Number of births				Prevalence (%)		
	Males	Females	Unknown	Total	Males	Females	Unknown	Total	Males	Females	Total[a]
1948–1958	140	102	3	245(34)	14,440	13,429	22	27,891	0.97	0.76	0.88(0.12)
1959–1969	184	176	2	362(59)	20,919	19,768	28	40,715	0.88	0.89	0.89(0.14)
1970–1980	273	201	8	482(85)	20,526	18,945	35	39,506	1.33	1.06	1.22(0.22)
1981–1990	188	140	1	329(48)	12,361	11,520	3	23,884	1.52	1.22	1.38(0.20)
Total	785	619	14	1,418(226)	68,246	63,662	88	131,996	1.15	0.97	1.07(0.17)

[a] Including sex unknown. Numbers in parentheses are multiply malformed cases and the prevalence.

Table 3. Secular changes in the prevalence[a] of selected congenital malformations, 1948–1990.

Malformations	1948–1958		1959–1969		1970–1980		1981–1990	
	No.	Pre.	No.	Pre.	No.	Pre.	No.	Pre.
Anencephaly	27	9.68	22	5.40	28	7.09	17	7.12
Spina bifida	7	2.51	5	1.23	16	4.05	3	1.26
Congenital hydrocephaly	3	1.08	14	3.44	6	1.52	6	2.51
Cleft lip	22	7.89	19	4.67	25	6.33	17	7.12
Cleft palate	16	5.74	19	4.67	17	4.30	8	3.35
Cleft lip and palate	10	3.59	20	4.91	36	9.11	20	8.37
Varus deformities of feet	68	24.38	91	22.35	99	25.06	51	21.35
Valgus deformities of feet	11	3.94	24	5.89	29	7.34	18	7.54
Atresia ani	6	2.15	11	2.70	13	3.29	8	3.35
Hermaphroditism	1	0.36	6	1.47	10	2.53	0	0
Hypospadias[b]	2	1.39	1	0.48	12	5.85	15	12.13
Hydrocele testis[b]	3	2.08	8	3.82	22	10.72	4	3.24
Polydactyly (finger)	19	6.81	39	9.58	30	7.59	15	6.28
Polydactyly (toe)	18	6.45	14	3.44	18	4.56	15	6.28
Syndactyly (finger)	2	0.72	7	1.72	15	3.80	7	2.93
Syndactyly (toe)	13	4.66	19	4.67	30	7.59	24	10.05
Down's syndrome	—	—	13	3.77[c]	22	5.57	25	10.47

[a] per 10,000 total births. [b] per 10,000 male births. [c] prevalence for 1961–1969.

the number of cases were divided by the total number of births during the period 1961–1990. Varus deformities of feet (23.4 per 10,000 births) occurred with highest prevalence, followed by polydactyly (finger) (7.8), anencephaly (7.1), cleft lip and palate (6.5), syndactyly (toe) (6.5), cleft lip (6.3), and Down's syndrome (6.1). The prevalence of each of polydactyly (finger) and anencephaly was high in both sexes. However, the prevalence of each of cleft palate, cleft lip and palate, syndactyly (finger), oligodactyly (finger), and hypoplastic auricle were significantly higher in males than in females.

Twins

Table 5 presents data on twins with malformations among 1,754 twin individuals. The prevalence of malformations was 1.20% (21/1,754) among all twins, and 1.46% (13/890) among male twins and 0.94% (8/853) among female twins. The difference between sexes is not significant. The prevalences for like-sexed and unlike-sexed twins were 1.23% (18/1,462) and 1.09% (3/276), respectively. There were no malformations among the triplets and quadruplets in the study. The prevalence of malformed infants among single births (1.07%) did not differ significantly from that among twins. The prevalence of anencephaly in twins was

Table 4. Numbers and prevalence of selected malformations, 1948–1990.

Malformation	Number of malformations			Prevalence (10^{-4})		
	Males	Females	Total[a]	Males	Females	Total[a]
Anencephaly	52	42	94	7.61	6.60	7.12
Spina bifida	11	20	31	1.61	3.14	2.35
Congenital hydrocephaly	15	13	29	2.20	2.04	2.20
Cleft lip	49	34	83	7.18	5.34	6.29
Cleft palate	22	37	60	3.22	5.81	4.55
Cleft lip and palate	59	27	86	8.65	4.24	6.52
Polydactyly (finger)	57	45	103	8.35	7.07	7.80
Syndactyly (finger)	22	9	31	3.22	1.41	2.35
Oligodactyly (finger)	11	3	14	1.61	0.47	1.06
Polydactyly (toe)	35	27	65	5.13	4.24	4.92
Syndactyly (toe)	46	40	86	6.74	6.28	6.52
Oligodactyly (toe)	4	3	9	0.59	0.47	0.68
Atresia ani	21	14	38	3.08	2.20	2.88
Hermaphroditism	11	4	17	1.61	0.63	1.29
Hypospadias	29	0	30	4.25	—	4.46
Hydrocele testis	37	0	37	5.42	—	5.42
Umbilical hernia	11	12	24	1.62	1.88	1.82
Super umbilical hernia	9	7	16	1.32	1.10	1.21
Hypoplastic auricle	23	6	29	3.37	0.94	2.20
Meatal stenosis	10	3	13	1.46	0.47	0.98
Diaphragmatic hernia	2	5	9	0.29	0.79	0.68
Down's syndrome[b]	28	32	60	5.54	6.78	6.13

[a] Including sex unknown. [b] The period from 1961 to 1990.

0.23% (4/1,754). There were two sets of conjoined twins among 131,102 deliveries, giving a crude rate of 15 per million deliveries.

DISCUSSION

Mitani and Kitamura (1968) conducted a longitudinal study of congenital malformations occurring at a hospital in Tokyo during the period 1922–1967; the overall prevalence was 0.83% and the prevalence decreased slightly by the year cohort. According to above authors, the prevalence was 0.79% during the priod 1948–1957, where data on congenital dislocation of the hip, accessory ear, and many kinds of congenital anomalies of heart were included. In the present study, although these malformations were excluded; the prevalence was higher (0.88%

Table 5. Twins with malformations among 1,754 twin individuals, 1948– 1990.

Malformations	Year of birth	Sex of twins
Anencephaly	1954	MM[a]
Anencephaly	1958	FF[a]
Anencephaly	1961	MM[a]
Anencephaly	1989	FF[a]
Cleft palate	1966	MM[a]
Cleft lip with cleft palate	1973	FM[a]
Hypospadias	1985	MM[a]
Polydactyly (finger)	1966	FF[b]
Polydactyly (toe)	1978	MF[a]
Oligodactyly (finger)	1986	MF[a]
Umbilical hernia	1969	FF[a]
Super umbilical hernia	1983	MM[a]
Atresia of external meatus	1976	MM[a]
Web neck	1980	MM[a]
Hydrocele testis	1982	MM[a]
Abnormal malposition of anus	1972	MM[a]
Conjoined twins	1971	MM
Conjoined twins	1971	FF

M: Male with malformation; F: Female with malformation. M[a]: Normal male; F[a]: Normal female. F[b]: Female with single umbilical artery.

for 1948–1958). According to Sumiyoshi *et al.* (1989), the overall prevalence of malformations increased year by year during the period 1972–1987, where the regression coefficient of prevalence on the year is significant at the 1% level. Similarly, in the present study, the overall prevalence increased significantly with the year cohort, where the lowest prevalence occurred in the earliest period (1948–1958) when data on Down's syndrome were not included. From Table 2 the ratio of overall prevalence during the period 1948–1969 (0.88%) to that during the period 1970–1990 (1.28%) was 1.5 and the difference is statistically significant at the 0.1% level. The corresponding ratios were 9.7 for hypospadias, 2.7 for syndactyly (finger), 2.5 for hydrocele testis, 2.0 for cleft lip and palate, 2.0 for Down's syndrome, and 1.8 for syndactyly (toe) (from Table 3). The prevalences for these congenital malformations were significantly higher in the latter period than in the former period. One of authors (HY) had been the director of St. Barnabas' Hospital during the period 1960–1986. However, he can't explain why the prevalence of hypospadias increased 9.7 times. On the other hand, the prevalences of Down's syndrome were 3.8 per 10,000 births for 1961–1969, 5.6 for 1970–1980, and 10.5 for 1981–1990. The corresponding values were 8.9 in Tottori Prefecture during

the period 1974–1983 (Takeshita *et al.*, 1984) and 9.9 in Kanagawa Prefecture during the period 1981–1983 (Kuroki and Konishi, 1984). Therefore there is good agreement among the present study during the period 1981–1990 and other Japanese studies for the period of the past 16 years. Then it seems that ascertainment of Down's syndrome had increased since 1970.

The prevalence of cleft palate decreased significantly with the year cohort (Table 3). According to Schull (1958), the rates of parental consanguineous marriages were higher in the patients of cleft palate than in the general population. On the other hand, the rates of consanguineous marriages gradually decreased in the period of 36 years from 1947 in Japan (Imaizumi *et al.*, 1975; Imaizumi, 1986). Then declining prevalence of cleft palate may be related to the declining rate of consanguineous marriages.

In the present study, the overall prevalence of malformations was 1.28% during the period 1970–1990. This can be compared with the prevalences of 1.21% in Tottori Prefecture during the period 1974–1983 (Takeshita *et al.*, 1984), 1.48% in Tokyo during the period 1979–1988 (Kato *et al.*, 1990), 1.14% in Kanagawa Prefecture during the period 1981–1983 (Kuroki and Konishi, 1984), and 0.82% in the whole of Japan during the period 1972–1987 (Sumiyoshi *et al.*, 1989). In the latter two studies, only 48 and 76 types of malformations were examined, respectively. This fact explains the low prevalences recorded in these studies.

In the present study, the prevalence was higher in twins (1.20%) than singletons (1.07%), but the difference is not significant. According to Little and Bryan (1988), malformations occur more frequently among twins than in singletons in most studies. In the present study, the prevalence of anencephaly (per 10,000 births) was 6.9 in singletons and 22.8 in twins, and the difference is significant at the 5% level. The corresponding values were 6.1 (1,626/2,658,357) and 9.1 (49/53,811), respectively, in a summary of data from several countries (Little and Bryan, 1988).

According to the Japanese Vital Statistics, the prevalence of conjoined twins was 10 per million births (Imaizumi, 1988). whereas the value was 15 per million births in the present study. According to Kato *et al.* (1990), the corresponding value was 19 in Tokyo Metropolitan Hospitals during the period 1979–1988. Edmonds and Layde (1982) estimated the values between 1:30,000 and 1:100,000 births in a summary of data from several countries.

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Appendix. Number of each type of malformation by sex

Malformation	Males	Females	Unknown	Total
I. Central Nervous System Malformations and Related Condition				
Anencephaly	52	42	0	94
Spina bifida	11	20	0	31
Congenital hydrocephaly	15	13	1	29
Microcephaly	3	5	0	8
Arhinencephaly	0	1	0	1
Cebocephaly	1	0	0	1
Meningocele	0	2	0	2
Cranium bifidum	3	2	0	5
Encephalocele	1	0	0	1
Pilonidal sinus	4	0	0	4
II. Musculoskeletal Abnormalities				
Varus deformities of feet	156	152	1	309
Polydactyly:				
Fingers	57	45	1	103
Toes	35	27	3	65
Syndactyly:				
Fingers	22	9	0	31
Toes	46	40	0	86
Oligodactyly:				
Fingers	11	3	0	14
Toes	4	3	2	9
Split hand	1	0	0	1
Split foot	1	1	0	2
Cleft-hand and cleft-foot	0	1	0	1
Abnormal position of toes	1	3	0	4
Valgus deformities of feet	39	43	0	82
Absence or hypoplasia of limb or part:				
Maxilla	1	0	0	1
Mandible	2	3	0	5
Fingers	7	3	0	10
Toes	2	1	0	3
Hand	1	0	0	1
Arm	0	2	0	2
Leg	1	1	0	2
Camptodactyly	5	1	0	6
Congenital clasped thumb	0	3	0	3
Inability to fingers contraction and extension	0	1	0	1
Femur defect	0	1	0	1
Absence of radial component	0	1	0	1
Radial ray defect	1	0	0	1
Talipes calcaneus	0	1	0	1
Arthrogryposis multiplex	1	0	0	1
Absence of skull bone	1	2	0	3
Absesce of frontal bone	1	1	0	2
Anomalies of spine	2	0	0	2
Vertebrae absence	1	0	0	1

Appendix (continue)

Malformations	Males	Females	Unknown	Total
Malformations of vertebral colum	0	1	0	1
Abnormal cuvature of vertebral column	1	0	0	1
Lumbar anomalies	0	1	0	1
Rib anomalies	0	1	0	1
Joint anomaly of finger	1	0	0	1
Joint anomaly of the limbs	1	0	0	1
Joint anomaly of knee	0	2	0	2
Knee disturbance of full extent	1	0	0	1
Ankylosis	0	1	0	1
Short limbs	2	4	0	6
Phocomelia	1	0	0	1
Amelia of upper and lower limbs	1	0	0	1
Anomaly of upper limb	1	0	0	1
Anomaly of lower limb	1	0	0	1
Anomaly of upper and lower limbs	3	2	0	5
Paralyzed humerus	1	0	0	1
Short feet	0	1	0	1
Macrodactyly(finger)	1	0	0	1
Macrodactyly(toe)	2	0	0	2
Arachnodactyly	1	0	0	1
Deformed wrists	1	0	0	1
Hallux varus	0	1	0	1
Anonychia of fingers or toes	3	2	0	5
Shortening of thigh	1	0	0	1
Abnormal thigh	1	0	0	1
Abnormal fingers	7	4	0	11
Abnormal toes	1	3	0	4
Abnormal hand	1	1	1	3
Abnormal foot	1	0	0	1
Abnormal forearm	1	0	0	1
Humerus varus	1	0	0	1
Congenital clubhand	1	1	0	2
Genu valgum	1	5	0	6
Acampsia	0	1	0	1
Pes adductus	0	1	0	1
Contracture of the wrist	1	0	0	1
Torticollis	5	2	0	7
Webbed neck	4	4	0	8
III. Genitourinary Conditions				
Retentio testis	10	0	0	10
Descensus testis	4	0	0	4
Urethroatresia	3	1	0	4
Hypospadias	29	0	1	30
Atresia ani	21	14	3	38
Hermaphroditism	11	4	2	17
Hydrocele testis	37	0	0	37
Scrotal hernia	2	0	0	2
Hypogenitalism	6	1	0	7
Anomalies of genital organs	6	1	1	8
Penile hypospadias	1	0	0	1
Small penis	4	0	0	4

Appendix (continue)

Malformations	Males	Females	Unknown	Total
Penile hydrops	1	0	0	1
Cystic kidney	0	1	0	1
Atresia hymenalis	0	1	0	1
Clitoral hypertrophy	0	1	0	1
Bifid scrotum	1	0	0	1
Dysplasia of external genital skin	0	1	0	1
Vaginal cyst	0	1	0	1
Ambiguous external genitalia	1	0	0	1
External genital cyst	1	0	0	1
Abnormal anus	3	0	0	3
Absence of ventral foreskin	1	0	0	1
IV. Eye Conditions				
Anophthalmia	3	2	0	5
Cyclops	1	3	0	4
Exophthalmos	1	2	0	3
Congenital cataract	0	2	0	2
Microphthalmia	3	1	0	4
Buphthalmos	1	0	0	1
Corneal opacity	0	2	0	2
Microcornea	0	1	0	1
Ankyloblepharon	2	1	0	3
Blepharophimosis	1	1	0	2
Hypertelorism	1	0	0	1
V. Ear Conditions				
Hypoplasia auricle	24	7	0	31
Atresia of external auditory canal	10	3	0	13
Atresia of auditory meatus	5	0	0	5
Absence of external auditory canal	2	1	0	3
Absence of pinna	0	3	0	3
Absence of auditory meatus	0	1	0	1
Microtia	0	1	0	1
Low set ears	3	3	0	6
Preauricular fistula	6	5	0	11
Deformed pinna	1	0	0	1
VI. Upper Respiratory Tract and Mouth Conditions				
Cleft lip	49	34	0	83
Cleft palate	22	37	1	60
Cleft lip with cleft palate	59	27	0	86
Micrognathia	0	1	0	1
Anterior nasal atresia	1	1	0	2
Absence of nasal cartilage	1	0	0	1
Saddle nose	0	1	0	1
Arhinia	1	0	0	1
Abnormal nose	1	1	0	2
Anomaly of oral region	2	0	0	2
VII. Thoracic Abnormalities				
Diaphragmatic hernia	2	5	2	9
VIII. Cardiovascula Conditions				
Patent ductus arteriosus	1	0	0	1
Tetralogy of Fallot	1	0	0	1
Atrial septal defects	0	1	0	1

Appendix (continue)

Malformations	Males	Females	Unknown	Total
Insufficiency of aortic	1	0	0	1
Single umbilical artery	2	2	0	4
IX. Alimentary Tract Malformations				
Umbilical hernia	11	12	1	24
Supra umbilical hernia	9	7	0	16
Esophageal atresia	6	1	0	7
Inguinal hernia	3	4	0	7
Ventral hernia	2	1	0	3
Gastroschisis	4	3	1	8
Intestinal atresia	3	0	0	3
Congenital atresia of intestine	1	0	0	1
Small intestine	0	1	0	1
Malrotation	0	1	0	1
Duodenal atresia	1	0	0	1
Duodenal stenosis	0	1	0	1
Rectal fistula	1	0	0	1
Umbilical malformations	0	1	0	1
Abnormal position of anus	1	1	0	2
Abnormal anus	3	0	0	3
Ectopia visceralis	4	0	1	5
Visceral hernia	2	1	0	3
X. Skin conditions and malformations				
Anonychia	3	1	0	4
Abnormal color of the nail	1	0	0	1
XI. Syndromes				
Down syndrome	28	32	0	60
Pierre Robin syndrome	0	2	0	2
Apert syndrome	1	0	0	1
Ellis van Creveld syndrome	0	1	0	1
Marfan's syndrome	1	0	0	1
Potter's syndrome	1	0	0	1
Leroy's syndrome	1	0	0	1
Cornelia De Lange syndrome	1	2	0	3
13 Trisomy	1	0	0	1
XII. Other				
Chondrodysplasia	6	3	0	9
Thrombocytopenia	0	1	0	1
Factor VIII deficiency	0	1	0	1
Mucopolysacharidosis	1	0	0	1
Congenital ichthyosis	1	0	0	1
Conjoined twins	1	1	0	2
Hydrops foetalis	0	4	0	4
Facial fissure	0	1	0	1
Facial anomalies	2	0	0	2
Defects of scalp skin	9	5	0	14