128. A Chromosomal Study on the 56 Cases of Cerebral Palsy

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Cerebral palsy is defined as any nonprogressive central motor deficit by the disease of brain, mainly motor center. The causal brain disease occurs at prenatal, perinatal, or postnatal period. In order to know the proportion of the chromosome abnormalities among the prenatal causes of the cerebral palsy, we made a chromosome survey on the patients who suffered from the cerebral palsy. This is a product of the direct combination-work undertaken among cytogeneticists, psychiatrists, and pediatrician. During a period from April 1982 to the end of May 1985, 56 cases with cerebral palsy, who were admitted in Hiroshima Prefectural Handicapped Children's Hospital, Wakaba Ryoikuen and in Kamo National Sanatorium, were collected for the chromosome research.

The chromosome slides were prepared following the standard blood culture procedures. The karyotype analyses were made with the application of the conventional Giemsa staining, and G- and C-banding differential staining.

Results and remarks. Table I showed the results of the clinical examinations and chromosomal findings. All patients had mental retardation; idiot, dyskinesia, difficulty or inability in walking, muscular atrophy, palsy or hemiplegia of extremities, and speech disorders. Then the above mentioned symptoms were excluded from Table I.

Fifty two cases among fifty six patients were found to have normal karyotypes, but the remaining four cases had abnormal karyotypes, being 7.1% in incidence.

They were represented by two cases of an inversion of no. 9 (inv(9)(p11q13) and inv(9)(p13q21)), one case of triplo-X (XXX), and one case of Turner's syndrome (X/XX/Xr(X)). The details of these cases were mentioned in the former papers (Kadotani *et al.* 1985 a, b; Kanata *et al.* 1985).

The causes of cerebral palsy fall into three main groups; prenatal, perinatal, and postnatal factors, as related to the period. Until now, the proportion of the prenatal causes has been small, and peri- and postnatal causes; asphyxia, forceps operation, placenta praevia, inertia uteri, prolonged labor, severe jaundice, meningitis, encephalitis, and so on, have occupied the large proportion; 80-90%. But from now, according to the advance of medicine, peri- and postnatal causes will decrease. The prenatal causes include chromosome abnormalities, hereditary diseases, intrauterine infection, and so on. The proportion of the chromosome abnormalities among the causes of cerebral palsy has remained uncertain. More data will be needed.

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Table I. Clinical and cytogenetic findings in 56 cases

Case no.	Age	Sex	Karyotype	Clinical features
1	8y.	F	46,XX	microcephaly, flat occiput, hypertelorism, misplasia and malinplantation in teeth, hypoplastic thumb, hypotonia, Lennox's syndrome
2	6y.	M	46,XY	Lecklinghausen's syndrome, dolichocephalia, low-set ears, misplasia and malinplantation in teeth, hypoplastic thumb, hypotonia, hypoplastic external genitalia, coarse hair, nevus (lt. femoral region)
3	9y.	F	46,XX	flat occiput, hypertelorism, hypoplastic thumb, club hands, club feet, calcaneous, hypotonia, seizure, hypothyroidism, kidney failure
4	26y.	M	46,XY	microcephaly, flat occiput, hypertelorism, micrognathia, misplasia and malinplantation in teeth, hypoplastic external genitalia, club hands, club feet, seizure
5	46y.	F	46,XX	coarse hair, flat occiput, club feet, seizure
6	11y.	F	46,XX	flat occiput, club feet, seizure
7	2y.	F	46,XX	flat occiput, strabismus, incurved index finger, seizure
8	34y.	M	46,XY	microcephaly, flat occiput, strabismus, hypoplastic external genitalia, incurved fingers, seizure
9	10y.	F	46,XX	head deformity, epicanthal fold, impaired vision, seizure, hypothyroidism, asthma
10	12y.	F	46,XX	head deformity, hypotonia, seizure
11	7y.	M	46,XY	flat occiput, ptosis of eyelid, hypoplastic external genitalia, hypotonia, hirsutism, seizure
12	44y.	F	46,XX	exophthalmus, strabismus, misplasia and malinplantation in teeth
13	26y.	M	46,XY	flat occiput, frontal bossing, hypoplastic external genitalia, incurved fingers, seizure
14	46y.	M	46,XY	congenital cataracta, seizure
15	23y.	M	46,XY	hypertelorism, incurved thumb
16	13y.	F	46,XX	microcephaly, flat occiput, misplasia and malinplantation in teeth, seizure
17	17y.	F	46,XX	seizure
18	18y.	M	46,XY	Sanfilippo's syndrome, saddle nose, seizure
19	13y.	M	46,XY	Sanfilippo's syndrome, saddle nose, nevus, high arched palate, varus
20	6y.	F	46,XX	blindness, seizure
21	15y.	M	46,XY	head deformity, strabismus, hypertonia, seizure
22	18y.	M	46,XY	ornitin transcarboamidase defect, hypertelorism, seizure
23	48y.	F	46,XX	hypertelorism, saddle nose, broad nasal base, upward slanted palpebral fissure
24	5y.	F	46,XX	strabismus, behavior disorders
25	7y.	M	46,XY	flat occiput, strabismus, behavior disorders
26	8y.	M	46,XY	hypertelorism, strabismus, impaired vision, behavior disorders, seizure
27	2y.	M	46,XY	head deformity, strabismus, seizure
28	6y.	F	47,XXX	blindness, prominent occiput, saddle nose, cleft

Table I (continued)

Case no.	Age	Sex	Karyotype	Clinical features
				palate, misplasia and malinplantation in teeth, incurved thumb, seizure
29	16y.	F	46, XX	hypertelorism, strabismus, saddle nose, cleft palate, hypertonia, athetosis
30	32y.	M	46, XY	seizure
31	26y.	M	46, XY	hypertelorism, flexion contracture of wrist, hypertonia
32	7y.	M	46, XY	blindness, head deformity, strabismus, nystagmus, low-set ears, misplasia and malinplantation in teeth, seizure, murmur
33	7y.	F	46, XY	hypertelorism, hypotonia, athetosis, seizure
34	6y.	M	46, XY	hypertelorism, upward slanted palpebral fissure, strabismus, saddle nose, hypotonia, Lennox's syndrome
35	7y.	M	46, XY	Lowe's syndrome, congenital cataracta, congenital glaucoma, flat occiput, upward slanted palpebral fissure, hypertelorism, epicanthus, misplasia and malinplantation in teeth, seizure
36	42y.	F	46, XX, inv (9) (p11q13)	hypertelorism, strabismus, upward slanted palpebral fissure, impaired vision, seizure
37	7y.	M	46, XY	misplasia and malinplantation in teeth
38	1y.	F	46, XX	Cornelia de Langé's syndrome, narrow high arched palate, impaired vision, micrognathia
39	10y.	F	46, XX	head deformity, hypertelorism, athetosis, seizure
40	1y.	F	46, XX	inverted epicanthus
41	10y.	F	46, XX	flat occiput, seizure
42	15y.	F	46, XX	low nasal bridge, cleft lip, seizure, behavior disorders
43	14y.	M	46, XY	cataracta, impaired vision, ataxia, behavior disorders
44	12y.	M	46, XY	flat occiput, club feet, club hands, seizure
45	17y.	F	46, XX	microcephaly, upward slanted palpebral fissure, seizure
46	13y.	M	46, XY	dolichocephaly, nystagmus, equino varus, seizure
47	18y.	M	46, XY	ataxia, seizure
48	21y.	F	45, X/46, XX/46, Xr(X)	short stature, microcephaly, hypertelorism, misplasia and malinplantation in teeth, hypogonadism, hypoplastic external genitalia, equino varus, hypoplasia of fourth toes at both, seizure
49	16y.	M	46, XY	blue sclera, impaired vision, seizure
50	34y.	F	46, XX	tuberous sclerosis, impaired vision, equino varus, hypertonia, adenoma sebaceum
51	13y.	M	46, XY	impaired vision, seizure
52	10y.	F	46, XX, inv (9) (p13q21)	micrognathia, seizure
53	18y.	F	46, XX	oxycephaly, seizure
54	12y.	F	46, XX	flat occiput, seizure
55	27y.	M	46, XY	microcephaly, seizure
56	15y.	F	46, XX	exophthalmus, ptosis of eyelid, saddle nose, seizure

All patients had mental retardation; idiot, dyskinesia, difficulty or inability in walking, muscular atrophy, palsy or hemiplegia of extremities, and speech disorders.

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