**84. Cytogenetic Data of 92 Cases Obtained in the
Kojika-Gakuen, an Institution for Severe
Physically and Mentally
Retarded Patients**

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Since 1978, cytogenetic information has been presented in 205 cases of congenital defectives and their relatives derived from genetic counseling at an institution for the congenital defectives.***) Additional data were accumulated in 92 cases from the Kojika-Gakuen, an institution for severe physically and mentally retarded patients. The present paper describes chromosomal findings in those cases in the form of summary together with their clinical features. This is a product of the combination-work of the Kojika-Gakuen and the Kadotani Medical Research Foundation.

According to the standard blood culture technique, chromosomal studies were carried out in 92 defectives and their relatives (for detail, refer to Table I). In each case, chromosome counts were made in 50 or more cells, and at least 10 cells with the modal chromosome-number were karyotyped. In cases with abnormal karyotypes, over 10 cells were analysed following the G- and Q-banding procedures.

Results and remarks. Table I shows the results of clinical examinations and chromosomal observations in 92 cases so far studied. Seventy among them were found to possess a normal karyotype, leaving 22 cases showing abnormal karyotypes, being 24% in incidence. They were represented by one case of triple X; two cases of the long Y; four cases of the short Y; two cases of the elongated short arm of no. 22; one case of the elongated satellite of no. 21; one case of the deleted short arm of no. 21; one case of trisomy 21; one case of the elongated short arm of D chromosome; one case of the elongated short arm of no. 15; one case of elongated satellite of no. 14; three cases of the elongated short arm of no. 13; one case of a mosaicism with ring no. 9 (46,XX/46,XX,r(9)(p24q34)); one case of the ab-

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^{***)} Katano *et al.*: Proc. Japan Acad., vol. 54B, pp. 111-115, 1978; vol. 56B, pp. 263-267, 1980.

Table I. Clinical notes and chromosomal findings of 92 cases from the Kojika-Gakuen for mentally retarded and physically defected patients

Case no.	Age	Sex	Karyotype	Clinical feature
1	6 y.	F	46,XX	slow growth, malnutrition, flat occiput, strabismus, shield chest, palsy of extremity, ataxia, idiot
2*	9 y.	F	46,XX,13p+	slow growth, malnutrition, microcephaly, ataxia, disturbance of gaits, idiot
3*	38 y.	M	46,XY,13p+	father of the case no. 2
4	34 y.	F	46,XX	mother of the case no. 2
5*	3 y.	M	46,XY,13p+	younger brother of the case no. 2
6	10 y.	F	46,XX	slow growth, malnutrition, flat occiput, round face, hypertelorism, upward slanted palpebral fissure, dislocation of hip, recurrent cramp, ataxia, idiot
7*	7 y.	F	46,XX,Dp+	slow growth, malnutrition, strabismus, atrial septal defect, palsy of extremity, hypotonicity, ataxia, idiot
8	8 y.	M	46,XY	low birth weight, spina bifida, clubfoot, valgus, palsy of extremity, athetotic movement, hypertonicity, recurrent cramp, ataxia, dysbasis, idiot
9	31 y.	F	46,XX	slow growth, malnutrition, round face, mask-like facies, palsy of extremity, hypertonicity, ataxia, disturbance of gaits, idiot
10	9 y.	M	46,XY	slow growth, malnutrition, short neck, round face, weak sight, palsy of extremity, muscular atrophy, ataxia, disturbance of gaits, idiot
11*	29 y.	F	46,XXX	slow growth, malnutrition, short neck, round face, cleft lip, teeth anomaly and irregular placement, atresia ani and vulvae, rocker bottom, palsy of extremity, ataxia, disturbance of gaits, idiot
12	14 y.	M	46,XY	flat occiput, strabismus, incomplete development of external genitalia, small penis, palsy of extremity, hypertonicity, ataxia, disturbance of gaits, idiot
13*	17 y.	M	46,XY,22p+	slow growth, clubhand, clubfoot, palsy of extremity, hypertonicity, recurrent cramp, ataxia, dysbasia, idiot
14*	48 y.	F	46,XX,22p+	mother of the case no. 13
15	11 y.	F	46,XX	younger sister of the case no. 13
16*	12 y.	F	46,XX,dir dup(3)(q22→q27)	low birth weight, sparse scalp hair, flat occiput, mask-like facies, recurrent cramp, ataxia, disturbance of gaits, idiot
17	34 y.	M	46,XY	father of the case no. 16
18	36 y.	F	46,XX	mother of the case no. 16
19	8 y.	F	46,XX	slow growth, malnutrition, low birth weight, flat occiput, weak sight, palsy of extremity, muscular atrophy, ataxia, disturbance of gaits, idiot
20	5 y.	F	46,XX	slow growth, malnutrition, low birth weight, nystagmus, pigeon breast, low-leg, palsy of extremity, hypertonicity, recurrent cramp, ataxia, disturbance of gaits, idiot
21	8 y.	M	46,XY	slow growth, malnutrition, short neck, round face, upward slanted palpebral fissure, teeth anomaly and irregular placement, clubhand, clubfoot, hypertonicity, hyperreflex of the tendon, recurrent cramp, ataxia, dysbasia, idiot
22	10 y.	M	46,XY	dolichocephalia, palsy of extremity, ataxia, disturbance of gaits, imbecile
23	34 y.	F	46,XX	slow growth, malnutrition, flat occiput, degeneration of retinal pigment, weak sight, saddle nose, low voice, teeth anomaly and irregular placement, palsy of extremity, muscular atrophy, recurrent cramp, ataxia, disturbance of gaits, debility
24	9 y.	M	46,XY	slow growth, malnutrition, hydrocephaly, frontal bossing, teeth anomaly and irregular placement, short neck, clubfoot, clubhand, incomplete development of internal and external genitalia, palsy of extremity, hypertonicity, recurrent cramp, ataxia, disturbance of gaits, idiot
25	6 y.	M	46,XY	slow growth, malnutrition, flat occiput, clubfoot, equinus, recurrent cramp, ataxia, disturbance of gaits, idiot
26	6 y.	F	46,XX	slow growth, malnutrition, flat occiput, palsy of extremity, hypertonicity, recurrent cramp, ataxia, disturbance of gaits, idiot
27	9 y.	F	46,XX	flat occiput, weak sight, palsy of extremity, ataxia, disturbance of gaits, idiot

Table I (Continued)

Case no.	Age	Sex	Karyotype	Clinical feature
28	25 y.	M	46,XY	saddle nose, low voice, clubfoot, equinus, rocker bottom, palsy of extremity, hypertonicity, malformed auricle (right), ataxia, disturbance of gaits, debility
29	7 y.	M	46,XY	microcephaly, oxycephaly, flat occiput, ataxia, disturbance of gaits, idiot
30	20 y.	M	46,XY	flat occiput teeth anomaly and irregular placement, clubhand, clubfoot, calcaneus, rocker bottom, palsy of extremity, muscle atrophy, recurrent cramp, ataxia, disturbance of gaits, idiot
31	24 y.	M	46,XY	macroglossia, clubfoot, equinovarus, dislocation of hip, palsy of extremity, ataxia, disturbance of gaits, idiot
32	11 y.	F	46,XX	flat occiput, strabismus, palsy of extremity, muscular atrophy, ataxia, disturbance of gaits, idiot
33	16 y.	M	46,XY	sparse scalp hair, dolichocephalia, flat occiput, frontal bossing, upward slanted palpebral fissure, exophthalmus, nystagmus, weak sight, palsy of extremity, muscular atrophy, ataxia, disturbance of gaits, idiot
34	49 y.	M	46,XY	microcephaly, cataract, weak sight, dysplasia of right I. finger, ataxia, disturbance of gaits, imbecile
35*	15 y.	M	46,XYq-	clubhand, clubfoot, equinus, palsy of extremity, hypertonicity, ataxia, disturbance of gaits
36	21 y.	M	46,XY	sparse scalp hair, microcephaly, flat occiput, short neck, protrusion of labium inferius, broad finger and toe, clubhand, clubfoot, equinus, rocker bottom, palsy of extremity, recurrent cramp, ataxia, disturbance of gaits, idiot
37	10 y.	M	46,XY	slow growth, flat occiput, clubfoot, palsy of extremity, ataxia, disturbance of gaits, idiot
38*	14 y.	M	46,XYq+	short stature, microcephaly, clubfoot, equinus, palsy of extremity, ataxia, disturbance of gaits, idiot
39	8 y.	F	46,XX	clubfoot, equinus, palsy of extremity, recurrent cramp, ataxia, disturbance of gaits, idiot
40	19 y.	F	46,XX	clubfoot, equinovarus, palsy of extremity, hypertonicity, ataxia, disturbance of gaits
41	48 y.	F	46,XX	aged face, weak sight, equinus, rocker bottom, palsy of extremity, recurrent cramp, ataxia, disturbance of gaits, idiot
42	28 y.	F	46,XX	microcephaly, flat occiput, weak sight, teeth anomaly and irregular placement, clubfoot, palsy of extremity, muscular atrophy, ataxia, disturbance of gaits, idiot
43	21 y.	F	46,XX	flat occiput, flat broad facies, upward slanted palpebral fissure, low nasal bridge, palsy of extremity, recurrent cramp, ataxia, disturbance of gaits, idiot
44	16 y.	F	46,XX	saddle nose, palsy of extremity, ataxia, disturbance of gaits, idiot
45	14 y.	F	46,XX	exophthalmus, cyanosis of extremity, palsy of extremity, ataxia, disturbance of gaits, idiot
46	18 y.	F	46,XX	strabism, saddle nose, cyanosis of extremity, palsy of extremity, recurrent cramp, ataxia, disturbance of gaits, idiot
47	38 y.	M	46,XY	rocker bottom, palsy of extremity, ataxia, disturbance of gaits, imbecile
48	16 y.	F	46,XX	slow growth, malnutrition, dry skin, microcephaly, strabism, saddle nose, low nasal bridge, protrusion of labium inferius, incomplete development of internal and external genitalia, wide cleft between I. and II. toe
49	12 y.	M	46,XY	slow growth, malnutrition, short stature, short neck, incomplete development of external genitalia, rocker bottom, palsy of extremity, hypertonicity, ataxia, disturbance of gaits, imbecile
50	23 y.	F	46,XX	dry skin, short neck, round face, facial telangiectatic erythema, flat occiput, microphthalmus, blindness, saddle nose, teeth anomaly and irregular placement, rocker bottom, recurrent cramp, ataxia, idiot
51	15 y.	F	46,XX	malnutrition, clubfoot, equinus, palsy of extremity, hypertonicity, recurrent cramp, ataxia, disturbance of gaits, debility

Table I (Continued)

Case no.	Age	Sex	Karyotype	Clinical feature
52	37 y.	F	46,XX	short stature, low birth weight, hirsutism, microcephaly, short neck, teeth anomaly and irregular placement, rocker bottom, palsy of extremity, hypertonicity, ataxia, disturbance of gaits, idiot
53	28 y.	F	46,XX	short neck, protrusion of labium inferius, ataxia, disturbance of gaits, idiot
54	28 y.	F	46,XX	brachycephaly, short neck, facial telangiectatic erythema, round face, saddle nose, athetotic movement, ataxia, disturbance of gaits, idiot
55	11 y.	F	46,XX	mask-like facies, hypertelorism, upward slanted palpebral fissure, saddle nose, protrusion of labium inferius, dislocation of hip, hypotonicity, ataxia, disturbance of gaits, idiot
56*	8 y.	F	46,XX/46,XX,r(9)(p24q34)	slow growth, malnutrition, short stature, dry skin, hirsutism, microcephaly, trigonocephaly, long eyelashes, slight down-ward slanted palpebral fissure, mask-like facies, macroglossia, protruding tongue, malformed low set ear, teeth anomaly and irregular placement, webbed neck, wide-spaced nipple, joint hyperextensibility, palsy of extremity, reflex acceleration, arachnodactyly, repeated convulsion, ataxia, disturbance of gaits, idiot
57	22 y.	F	46,XX	slow growth, malnutrition, short stature, skin pigmentation, nevus vasculosus, dry skin, flat occiput, funnel breast, incomplete development of external genitalia, ataxia, idiot
58	39 y.	F	46,XX	slow growth, malnutrition, short neck, facial telangiectatic erythema, hypertelorism, saddle nose, low nasal bridge, protrusion of labium inferius, brachydactyly, teeth anomaly and irregular placement, V. finger clinodactyly, IV toe hypoplasia, ataxia, disturbance of gaits, idiot
59*	25 y.	M	46,XYq--	clubhand, clubfoot, equinus, palsy of extremity, hypertonicity, ataxia, disturbance of gaits, imbecile
60	22 y.	F	46,XX	facial telangiectatic erythema, idiot
61	25 y.	M	46,XY	dolichocephalia, flat occiput, facial telangiectatic erythema, cataract (right), incomplete development of external genitalia, vagus, ataxia, disturbance of gaits, idiot
62	27 y.	M	46,XY	slow growth, ataxia, idiot
63*	18 y.	M	46,XY,21p--	hirsutism, recurrent cramp, ataxia, disturbance of gaits, idiot
64	28 y.	F	46,XX	short neck, ataxia, disturbance of gaits, idiot
65	11 y.	M	46,XY	dry skin, hirsutism, flat occiput, strabismus (left), weak sight, recurrent cramp, ataxia, disturbance of gaits
66	36 y.	M	46,XY	slow growth, malnutrition, short stature, small cranium, older facies, prominent ears, clubhand, clubfoot, cavus, ataxia, disturbance of gaits, idiot
67	37 y.	M	46,XY	skin pigmentation, dry skin, short neck, round face, exophthalmus, macroglossia, wide cleft between I. and II. toe, idiot
68*	11 y.	M	47,XY,+21	slow growth, malnutrition, short stature, microcephaly, hypertelorism, strabismus, blepharophimosis, blepharitis, saddle nose, macroglossia, joint hyperextensibility, simian-line, ataxia, disturbance of gaits, idiot
69	22 y.	M	46,XY	albino, flat occiput, red hair, round face, protrusion of labium inferius, idiot
70	15 y.	F	46,XX	round face, saddle nose, low nasal bridge, teeth anomaly and irregular placement, thick lip, female like breast, recurrent cramp, ataxia, disturbance of gaits, imbecile
71	18 y.	M	46,XY	flat occiput, protrusion of labium inferius, teeth anomaly and irregular placement, hypodontia, pigeon breast, ataxia, disturbance of gaits, idiot
72	14 y.	M	46,XY	weak sight, idiot
73	34 y.	M	46,XY	dolichocephalia, flat occiput, aged facies, inner epicanthal fold, malformed auricle, prognathism, narrow high arched palate, ataxia, disturbance of gaits, idiot
74*	30 y.	M	46,XYq--	low birth weight, flat occiput, idiot
75	18 y.	F	46,XX	low birth weight, dry skin, microcephaly, flat occiput, protrusion of labium inferius, teeth anomaly and irregular placement, palsy of extremity, recurrent cramp, disturbance of gaits, idiot

Table I (Continued)

Case no.	Age	Sex	Karyotype	Clinical feature
76	14 y.	M	46,XY	short neck, round face, weak sight, idiot
77*	11 y.	M	46,XYq-	slow growth, teeth anomaly and irregular placement, thick lip, left hemiplegie, palsy of extremity, muscle atrophy, recurrent cramp, ataxia, disturbance of gaits, idiot
78*	36 y.	F	46,XX,21s+	dry skin, exophthalmus, malformed auricle, malformed limbs, recurrent cramp, ataxia, idiot
79*	16 y.	M	46,XY,15p+	short stature, macrocephaly, short neck, round face, ptosis of eyelid, small nose, teeth anomaly and irregular placement, short and sparse cilia, incomplete development of internal and external genitalia, small penis, short extremities, ataxia, disturbance of gaits, idiot
80*	30 y.	M	46,XY,14s+	skin pigmentation, exophthalmus, protrusion of labium inferius, teeth anomaly and irregular placement, idiot
81	28 y.	M	46,XY	cataract, blindness, idiot
82	19 y.	M	46,XY	varus, simianline, debility
83	27 y.	M	46,XY	hirsutism, microcephaly, flat occiput, blepharitis, blindness, prominent nasal bridge, teeth anomaly and irregular placement, ataxia, disturbance of gaits, idiot
84	9 y.	M	46,XY	microcephaly, brachycephaly, round face, protrusion of labium inferius, rocker bottom, palsy of extremity, ataxia, disturbance of gaits, idiot
85	27 y.	F	46,XX	dry skin, blepharophimosis, nose depressed, long II. toe, recurrent cramp, ataxia, disturbance of gaits, idiot
86*	12 y.	M	46,XY,der(1),ins(1;9)(q11;q12q13)mat	goose flesh, dry and hard skin, dolichocephalia, flat occiput, upward slanted palpebral fissure, blepharophimosis, weak sight, nose depressed, low nasal bridge, teeth anomaly and irregular placement, low set ear, incomplete development of internal and external genitalia, cryptorchism, small penis, clubfoot, varus, repeated convulsion, imbecile
87	34 y.	M	46,XY	father of the case no. 86
88*	30 y.	F	46,XX,ins(1;9)(q11;q12q13)	mother of the case no. 86, normal phenotype
89	29 y.	F	46,XX	hirsutism, round face, idiot
90	24 y.	M	46,XY	hirsutism, upward slanted palpebral fissure, saddle nose, macroglossia, clubfoot, palsy of leg, ataxia, disturbance of gaits, idiot
91*	11 y.	M	46,XYq+	low birth weight, dolichocephalia, dry skin, facial telangiectatic erythema, nose depressed, incomplete development of internal and external genitalia, short limbs, brachydactyly, debility
92	12 y.	F	46,XX	idiot

Cases with asterisks are chromosomally abnormal.

normal long arm of no. 3 resulting the partial duplication of the long arm of no. 3 (q22→q27) (46,XX,dir dup(3) (q22→q27)) ; one case of a partial trisomy for the long arm of no. 9 (46,XY,der(1),ins(1;9)(q11;q12q13)mat) ; and one case showing a direct insertion of the long arm of no. 9 into the long arm of no. 1 (46,XX,ins(1;9)(q11;q12q13)).

Among the abnormalities, one case of the partial duplication of the long arm of no. 3 (case no. 16) seemed to be of sporadic origin, since the parents of this case were chromosomally normal.*¹) Details of the case with a mosaicism with ring no. 9 (case no. 56) were reported by Kadotani *et al.***²) Partial trisomy for the long arm of no. 9

*¹) Kadotani *et al.*: Proc. Japan Acad., vol. 55B, pp. 431-435, 1979.

**²) Kadotani *et al.*: Proc. Japan Acad., vol. 56B, pp. 85-88, 1980.

(case no. 86) was transmitted through the maternal line, since the mother of this case (case no. 88) was a carrier of the direct insertion of a long arm of no. 9 into a long arm of no. 1.*)

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*) Kadotani *et al.*: Proc. Japan Acad., vol. 56B, pp. 157-161, 1980.