7. A Chromosome Study on the 91 Cases of the Mentally Retarded Men

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A chromosomal survey was done in the patients in Roppo-Gakuen, which was an establishment for the mentally retarded men, during a period from February 1981 to February 1983, 91 cases were collected for the chromosomal study.

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

Results and remarks. Table I showed the results of the clinical examinations and chromosomal findings. All patients had mental retardation; imbecile.

Eighteen cases out of ninety one cases were found to possess the abnormal karyotypes, being 19.8% in incidence. But the remaining seventy-three cases had the normal karyotype. And thirteen cases out of the abnormal eighteen cases were Down's syndrome, being 72.2% in incidence (Table I). The abnormalities were found at a little low level in incidence to those of the former survey; 23.5% in incidence in congenital defectives,¹⁾ 26.2% in incidence in the patients such as a cerebral palsy and seizure.²⁾

They were represented by eighteen cases; one case of a reciprocal translocation between no. 7 and no. 14 chromosomes (46,XY,rcp(7;14)(q11;q22)), one case of a deletion of no. 10 chromosome (46,XY,del(10)(p14)), one case of the translocation between no. 13 and no. 14 chromosomes (45,XY,-13,-14,+t(13;14)(p11;p11)), one case of the ring chromosome of no. 18 (46,XX,r(18)(p11;q23)), eleven cases of Down's syndrome having trisomy no. 21 chromosomes (47,XX or XY,+21), two cases of translocational Down's syndrome (46,XX,t(14;21)), and one case having a very small marker chromosome (47,XX,+mar).

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References

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- 2) Kadotani, T. *et al.* (1984): *ibid.*, 60B, 129-130.

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Table I. Clinical and cytogenetic findings in 91 cases

Case no.	Age	Sex	Karyotype	Clinical features
1.	33y.	F	46, XX	dilatation of the heart, anemia, adrenal hypertension, short stature, hypertelorism, low nasal bridge, flat nasal base, small nose, flat breasts, hypoplastic thumb
2.	32y.	F	46, XX	hirsutism
3.	38y.	F	46, XX	vagabondism
4.	37y.	F	47, XX, +21	Down's syndrome, short neck, flat facies, epicanthus, upward slanted palpebral fissure, low nasal bridge, saddle nose, flat nasal base, narrow high arched palate, hypoplastic thumb, V. finger clinodactyly
5.	23y.	F	46, XX	moon facies, flat nasal base, large nose, narrow high arched palate
6.	21y.	F	47, XX, +21	Down's syndrome, flat occiput, short neck, upward slanted palpebral fissure, epicanthus, low nasal bridge, saddle nose, flat nasal base, brachydactyly
7.	24y.	F	46, XX	narrow high arched palate
8.	24y.	F	46, XX	short stature, narrow high arched palate, misplasia and malimplantation in teeth, small thorax of left, hypoplastic thumb, arachnodactyly
9.	23y.	F	47, XX, +mar	brain edema, epilepsy, short stature, narrow high arched palate, nocturnal enuresis
10.	26y.	F	46, XX	slightly epilepsy, narrow high arched palate
11.	23y.	M	46, XY	normal feature
12.	34y.	F	46, XX	flat occiput, exophthalmus, epicanthus
13.	23y.	F	46, XX	flat occiput, enlargement of external genitalia
14.	33y.	M	46, XY	short stature, simianline of right hand
15.	22y.	M	46, XY	normal feature
16.	34y.	M	46, XY	normal feature
17.	29y.	M	46, XY	normal feature
18.	22y.	M	46, XY	normal feature
19.	38y.	F	46, XX	flat facies, double-eyelid, hypertelorism, ptosis of eyelid
20.	22y.	M	47, XY, +21	Down's syndrome, short stature, small head, flat occiput, moon facies, upward slanted palpebral fissure, epicanthus, low nasal bridge, flat nasal base, high arched palate, brachydactyly, hypotonia, simianline of both hands
21.	24y.	M	46, XY	flat and moon facies, V. finger clinodactyly, hypotonia, simianline of both hands
22.	40y.	M	46, XY	normal feature
23.	22y.	F	47, XX, +21	Down's syndrome, short stature, microcephaly, short and web neck, flat and moon facies, older facies, hypertelorism, upward slanted palpebral fissure, ptosis of eyelid, epicanthus, low set ears, malformed auricle, low nasal bridge, nose depressed, flat nasal base, high arched palate, V. finger clinodactyly, hypotonia, simianline of both hands

Table I (continued)

Case no.	Age	Sex	Karyotype	Clinical features
24.	19y.	M	46,XY	short stature, dolichocephalia, high arched palate
25.	35y.	M	46,XY	normal feature
26.	35y.	M	46,XY	normal feature
27.	34y.	M	46,XY	stubby finger
28.	22y.	M	46,XY	dolichocephalia, upward slanted palpebral fissure, hirsutism, high arched palate
29.	19y.	F	46,XX	double-eyelid, flat nasal base, large nose
30.	31y.	F	46,XX	normal feature
31.	24y.	F	46,XX	microcephaly, flat occiput, epicanthus, prominent nasal bridge, hypoplastic thumb
32.	19y.	F	46,XX	hirsutism, moon facies, hypertelorism, epicanthus, large nose, hypoplastic thumb, V. finger clinodactyly, hypertonicity
33.	12y.	M	47,XY, +21	Down's syndrome, flat occiput, short neck, flat forehead, moon facies, hypertelorism, upward slanted palpebral fissure, epicanthus, short and coarse eyelashes, low set ears, malformed auricle, protruding tongue, hypoplastic thumb, V. finger clinodactyly
34.	26y.	M	46,XY	microcephaly, hypertelorism, upward slanted palpebral fissure, prominent nasal bridge, involuntary movement, ataxia
35.	12y.	M	46,XY	macrocephaly, low set ears, prominent nasal bridge, V. finger clinodactyly, hypotonia
36.	20y.	F	46,XX	hirsutism, macrocephaly, moon facies, strabismus of left, flat nasal base, hypoplastic thumb, V. finger clinodactyly, ataxia
37.	7y.	M	46,XY, rep(7;14)(q11;q22)	sparse hair, scanty hair of the scalp, dolichocephalia, oxycephaly, flat occiput, hypertelorism, low set and large ears, low nasal bridge, flat nasal base, V. finger clinodactyly at both sides
38.	11y.	F	46,XX	tiny microcephaly, V. finger clinodactyly
39.	12y.	F	46,XX	macrocephaly, dolichocephalia, hypertelorism, epicanthus, low set and simple ears, flat nasal bridge
40.	17y.	F	46,XX	flat occiput, mask-like facies, hypertelorism, epicanthus, low set ears, flat nasal bridge, hypotonia, simianline of left hand
41.	18y.	F	47,XX, +21	mask-like facies, hypertelorism, upward slanted palpebral fissure, epicanthus, low set ears, low nasal bridge, flat nasal base, small nose, hypotonia, simianline of left, short limbs
42.	15y.	F	46,XX, r(18)(p11;q23)	flat nasal base, simianline of both hands
43.	17y.	F	46,XX	flat occiput, moon facies, epicanthus, saddle nose, flat nasal base, V. finger clinodactyly, simianline of both hands, hypotonia
44.	19y.	F	46,XX	flat occiput, hypotelorism, upward slanted palpebral fissure, epicanthus, saddle nose, flat nasal base, large alae nasi

Table I (continued)

Case no.	Age	Sex	Karyotype	Clinical features
45.	18y.	F	47, XX, +21	Down's syndrome, microcephaly, flat occiput, short neck, flat forehead, moon facies, upward slanted palpebral fissure, ptosis of eyelid, epicanthus, low set ears, flat nasal base, narrow high arched palate, hypoplastic thumb, V. finger clinodactyly, simianline of left hand
46.	21y.	F	46, XX	flat occiput, narrow high arched palate
47.	20y.	F	47, XX, +21	Down's syndrome, short stature, scanty hair of the scalp, microcephaly, flat occiput, lines of brow, upward slanted palpebral fissure, epicanthus, blepharophimosis, saddle nose, flat nasal base, high arched palate, V. finger clinodactyly, simianline of right, hypotonia
48.	16y.	F	46, XX	flat occiput, strabismus of both eyes, prominent nasal bridge, high arched palate
49.	20y.	F	46, XX, t(14; 21)	Down's syndrome, scanty hair of the scalp, microcephaly, flat occiput, upward slanted palpebral fissure, epicanthus, low set ears, flat nasal base
50.	21y.	F	47, XX, +21	Down's syndrome, microcephaly, flat occiput, short neck, upward slanted palpebral fissure, epicanthus, blepharophimosis, short and coarse eyelashes, low set ears, nose depressed, flat nasal base, small nose, high arched palate, prominent labium inferius, V. finger clinodactyly, simianlines of both hands, hypotonia
51.	22y.	F	46, XX, t(14; 21)	Down's syndrome, short stature, microcephaly, flat occiput, upward slanted palpebral fissure, epicanthus, short and coarse eyelashes, low set ears, flat nasal base, high arched palate, prominent labium inferius, brachydactyly, hypotonia
52.	20y.	F	46, XX	microcephaly, flat occiput, external strabismus
53.	13y.	F	46, XX	nose depressed
54.	17y.	F	46, XX	macrocephaly, hardness of hearing, speech disorders
55.	23y.	F	46, XX	short and coarse eyelashes, low nasal bridge, flat nasal base, high arched palate
56.	21y.	F	46, XX	macrocephaly, long eyelashes
57.	21y.	F	46, XX	microcephaly
58.	24y.	F	46, XX	normal feature
59.	18y.	F	46, XX	normal feature
60.	20y.	F	46, XX	hypertelorism, low nasal bridge, simianlines of both hands
61.	18y.	F	46, XX	dolichocephalia, low nasal bridge, prominent labium inferius, small labium
62.	14y.	M	46, XY	malformed auricle
63.	15y.	M	46, XY	hypertelorism, upward slanted palpebral fissure, blepharophimosis, flat nasal base, simianline of right
64.	16y.	M	46, XY	V. finger clinodactyly

Table I (continued)

Case no.	Age	Sex	Karyotype	Clinical features
65.	18y.	M	45, XY, -13, -14, +t(13; 14) (p11; p11)	macrocephaly, malformed auricle, high arched palate
66.	17y.	M	46, XY	hypotelorism, malformed auricle
67.	17y.	M	46, XY, del(10) (p14)	macrocephaly, flat occiput, epicanthus, microtia, low nasal bridge, flat nasal base
68.	16y.	M	46, XY	long hands, simianline of right
69.	16y.	M	46, XY	microcephaly, oxycephaly
70.	16y.	M	46, XY	microcephaly, oxycephaly, flat occiput, upward slanted palpebral fissure, epicanthus, large ear, arachnodactyly
71.	16y.	M	46, XY	long stature, flat occiput, hypotelorism
72.	17y.	M	46, XY	dolichocephalia, flat occiput, short neck, long fingers
73.	18y.	M	46, XY	flat occiput, low set ears, large nose
74.	18y.	M	46, XY	downward slanted palpebral fissure, malformed auricle, long fingers, hypotonia
75.	8y.	M	47, XY, +21	Down's syndrome, scanty hair of the scalp, microcephaly, flat occiput, short neck, flat facies, hypertelorism, upward slanted palpebral fissure, short and coarse eyelashes, low set ears, low nasal bridge, saddle nose, flat nasal base, small nose, brachydactyly, V. finger clinodactyly, simianlines of both hands
76.	9y.	M	46, XY	oxycephaly, head deformity, mouth opening, V. finger clinodactyly
77.	8y.	M	47, XY, +21	Down's syndrome, scanty hair of the scalp, microcephaly, flat occiput, short neck, moon facies, hypertelorism, upward slanted palpebral fissure, epicanthus, low set ears, malformed auricle, low nasal bridge, nose depressed, saddle nose, flat nasal base, small nose, mouth opening, brachydactyly, simianlines of both hands, hypotonia
78.	14y.	M	46, XY	dolichocephalia, downward slanted palpebral fissure, mouth opening, long fingers, simianline of left
79.	16y.	M	46, XY	macrocephaly, oxycephaly, short neck, flat facies, hypotelorism, epicanthus, short and coarse eyelashes, blepharitis, low set ears, malformed auricle, low nasal bridge, nose depressed, saddle nose, flat nasal base, large nose, mouth opening, protruding tongue, polydactyly
80.	10y.	M	46, XY	macrocephaly, oxycephaly, flat occiput, downward slanted palpebral fissure, low nasal bridge, saddle nose, large nose
81.	16y.	M	46, XY	macrocephaly, low set ears, malformed auricle, misplasia and malinplantation in teeth, epilepsy
82.	8y.	M	46, XY	hypertelorism, high arched palate
83.	15y.	M	46, XY	macrocephaly, moon facies, high arched palate

Table I (continued)

Case no.	Age	Sex	Karyotype	Clinical features
84.	14y.	M	46, XY	dolichocephalia, hypotelorism, large nose, high arched palate
85.	20y.	M	46, XY	hypotelorism, malformed auricle, large nose, high arched palate, simianline of right
86.	13y.	M	46, XY	dolichocephalia, epicanthus, prominent nasal bridge
87.	18y.	M	46, XY	microcephaly, head deformity, hypotelorism, epicanthus, malformed auricle, prominent nasal bridge, beaked nose, high arched palate
88.	8y.	M	46, XY	hypertelorism, epicanthus, low nasal bridge, nose depressed
89.	22y.	M	46, XY	microcephaly, epicanthus, low nasal bridge, nose depressed, large nose, idiot
90.	13y.	M	46, XY	scanty hair of the scalp, dolichocephalia, flat occiput, hypertelorism, epicanthus, low nasal bridge, large nose
91.	18y.	M	46, XY	high arched palate