- ⁶ Greig DM. Oxycephaly. *Edinburgh Med J* 1928;33:189-218.
- ⁷ Canki N. Syndactyly+polydactyly+triphthalangeal thumbs with tibial hypoplasia and nose anomaly in two generations: a new syndrome. *5th International Conference on Birth Defects*, Montreal, 1977.
- ⁸ Reber M. Un syndrome osseux peu commun associe une heptadactylic et une aplasie des tibiae. *J Genet Hum* 1967;16:15-39.
- ⁹ Pashayan H, Fraser FC, McIntyre M, Dunbar J. Bilateral aplasia of the tibiae, polydactyly and absent thumb in father and daughter. *J Bone Joint Surg (Br)* 1971;53:495-9.
- ¹⁰ Al-Awadi SA, Moussa MA, Naguib KK, et al. Consanguinity among the Kuwaiti population. *Clin Genet* 1985;27:483-6.
- ¹¹ Pauli R. Dominance and homozygosity in man. *Am J Med Genet* 1983;16:455-8.
- ¹² Hirschhorn K. Dominance and homozygosity in man. *Am J Med Genet* 1984;18:534.
- ¹³ Temtamy S, McKusick V. Polydactyly. In: *The genetics of hand malformations*. Birth Defects, vol XIV, No 3, New York: The National Foundation-March of Dimes, 1978:364-439.
- ¹⁴ McKusick VA. *Mendelian inheritance in man. Catalog of autosomal dominant, autosomal recessive and X-linked phenotypes*. 6th ed. Baltimore: Johns Hopkins University Press, 1983.

Correspondence and requests for reprints to Dr Kamal K Naguib, Kuwait Medical Genetics Centre Maternity Hospital, PO Box 4080, 13041 Safat Kuwait.

Duplication 14(q31→qter)

DAISY M CARR, KWEI JONES-QUARTEY*, MARGARITA V VARTANIAN, AND HELEN MOORE-KAPLAN

Cytogenetics Laboratory, Department of Pathology, Charles Drew/UCLA Medical School, Los Angeles, California 90059, USA.

SUMMARY A 29 year old black female with delayed development and multiple congenital anomalies showing a duplication of 14(q31→qter) is presented. Although clinical features associated with partial duplication of the distal long arm of chromosome 14 are variable, there are some features which are distinctive of this chromosomal abnormality. The clinical findings of 14 cases with duplication distal 14q are compared in an effort to define a recognisable syndrome.

Duplication of 14p, the centromeric region, and various segments of proximal 14q, has been observed in numerous patients with mental retardation and multiple congenital anomalies. Duplication of distal 14q, however, has been observed in only a few cases. The present paper reports a case of duplication 14(q31→qter).

Case report

A 29 year old black female was referred to the King-Drew Medical Center from a nursing home because of vomiting and weight loss of five to six months' duration. Weight loss was ascribed to decreased food

intake. There were no other gastrointestinal symptoms. The patient had had a diagnosis of encephalopathy due to unknown cause and defective fetal development with multiple congenital anomalies. Her IQ from a previous evaluation was at an 11 year old level. She had been born prematurely at 7½ months gestation, the third child of a mother described as being anaemic and having unspecified cardiac problems. The patient had a history of frequent upper respiratory infections and bilateral pneumonia. She had had extensive dental surgery for multiple caries and periodontitis.

Upon admission, the patient was afebrile with a blood pressure of 100/70. She was poorly nourished and underdeveloped with proportionately small

FIGURE Chromosome 14 pairs from the proband showing duplication of 14 (q31→qter).

*Present address: Department of Internal Medicine, Los Angeles County-USC Medical Center, Los Angeles, California 90033, USA.

Received for publication 22 April 1986.
Accepted for publication 24 April 1986.

TABLE Clinical features observed in 14 cases of duplication of distal 14q.

	Pfeiffer et al ¹	Fryns et al ⁴	Trunca and Opitz ¹⁰	Wyandt et al ¹¹	Bridgman and Butler ²	Geormaneanu et al ⁵	Sklower et al ⁹	Sklower et al ⁹	Sklower et al ⁹	Cohen et al ³	Nikolis et al ⁶	Romain et al ⁸	Atkin and Puth ¹	Present case
Duration of pregnancy (wk)				Term	35	F	5 mth	2000	+					
Sex	F	41	40	F	35	F	2-8 mth	1800	Term	39	Term	Term	40	30
Survival	3660	2-5 mth	1920	3070	2000	2070	2-8 mth	2070	M	1730	4 mth	M	F	F
Birth weight (g)		2500								8-5 mth	2250	2480	3800	29 y
Failure to thrive		+	+	+	+	+		+		+	+		+	+
Growth retardation		+	+	+	+	+		+		+	+		+	+
Motor retardation		+	+	+	+	+		+		+	+		+	+
Mental retardation		+	+	+	+	+		+		+	+		+	+
Hypotonia		+	+	+	+	+		+		+	+		+	+
Brachycephaly		+	+	+	+	+		+		+	+		+	+
Microcephaly		+	+	+	+	+		+		+	+		+	+
Facial asymmetry		+	+	+	+	+		+		+	+		+	+
Micrognathia		+	+	+	+	+		+		+	+		+	+
Epicanthus		+	+	+	+	+		+		+	+		+	+
Antimongoloid slant		+	+	+	+	+		+		+	+		+	+
Hypertelorism		+	+	+	+	+		+		+	+		+	+
Blindness		+	+	+	+	+		+		+	+		+	+
Prosis		+	+	+	+	+		+		+	+		+	+
Short palpebral fissures		+	+	+	+	+		+		+	+		+	+
Carp shaped mouth		+	+	+	+	+		+		+	+		+	+
Low set or malformed auricles		+	+	+	+	+		+		+	+		+	+
Anteverted nostrils		+	+	+	+	+		+		+	+		+	+
Deafness		+	+	+	+	+		+		+	+		+	+
Cardiovascular anomalies		+	+	+	+	+		+		+	+		+	+
Club feet		+	+	+	+	+		+		+	+		+	+
Digital anomalies		+	+	+	+	+		+		+	+		+	+
Hypogonadism		+	+	+	+	+		+		+	+		+	+
Undescended testes		+	+	+	+	+		+		+	+		+	+
Congenital glaucoma		+	+	+	+	+		+		+	+		+	+
Absence of parathyroids		+	+	+	+	+		+		+	+		+	+
High arched palate		+	+	+	+	+		+		+	+		+	+

head, trunk, and limbs. The skin was tight and hairless over the upper and lower limbs. There was hypertelorism and the fundi were normal. The external ears showed poorly developed tragi and helices. The large protruding tongue was in constant motion and the oral cavity revealed multiple filled teeth. The breasts were undeveloped with widely spaced nipples and the chest revealed bilateral rhonchi. The heart was unremarkable with no murmurs. There was voluntary abdominal guarding. The external genitalia revealed no lesions, but the vagina was small and not easily examined. Digital anomalies of the hands were observed but there was no simian crease. There were flexion contractures at the elbow, hip, and knee joints. Initial serum electrolytes were consistent with dehydration. The CBC was unremarkable and thyroid function tests were normal. The chest x ray showed bilateral pneumonic infiltrates thought to be consistent with aspiration pneumonia. A CT scan of the head revealed central cerebral atrophy with no mass lesions. An attempted upper gastrointestinal series was unsuccessful because of the patient's inability to cooperate. An oesophagogastrroduodenoscopy revealed hiatus hernia and reflux, but a normal oesophagus. The patient was discharged to the nursing home in a stable condition and died a few months later.

CYTOGENETIC STUDIES

Chromosomal analyses were performed on PHA stimulated peripheral lymphocytes using trypsin-Giemsa banding techniques. The karyotype was 46,XX,dup(14)(q31→qter) (figure). The parents of the proband were unavailable for chromosome analyses.

Discussion

Phenotypic heterogeneity has been observed in patients with partial duplication of various chromosomal segments. Unlike complete or mosaic trisomies, partial duplications of chromosome 14 are fairly well tolerated and the severity of the anomaly apparently depends on which segment of the long arm is involved. Patients in whom only a small proximal part of the chromosome is duplicated show minor malformations and mild mental deficiency. Whenever a larger or more distal part of the long

arm is involved, the malformations are more pronounced and the mental deficiency is severe.¹⁻¹¹

As seen in the table, clinical findings associated with partial duplication of distal 14q are variable but some features remain characteristic of this chromosomal rearrangement. The condition is marked by severe mental, motor, and growth retardation. The neurological function, as seen in the case reported here, seems to be profoundly affected. Other distinctive features shared by these patients include malformed or low set auricles, antimongoloid slant, hypertelorism, micrognathia, digital and cardiovascular anomalies, and low birth weight.

The secretarial assistance of Miss Jeraldine Brown is gratefully acknowledged.

References

- Atkin JF, Patil S. Duplication of the distal segment of 14q. *Am J Med Genet* 1983;16:357-66.
- Bridgman G, Butler LJ. A child trisomic for the distal part of chromosome 14q. *Arch Dis Child* 1980;55:474-7.
- Cohen MM, Charrow J, Balkin NE, Harris CJ. Partial trisomy 14 (q23→qter) via segregation of a 14/X translocation. *Am J Hum Genet* 1983;35:635-44.
- Fryns JP, Van Eygen M, Tanghe W, Van den Berghe H. Partial trisomy 14q due to familial t(14q-;11q+) translocation. *Hum Genet* 1977;37:105-10.
- Geormaneanu M, Geormaneanu C, Walter-Rosianu A, Papuc M. Trisomie 14q distale associee a l'agenesie du corps calleux et au tronc arteriel commun, due a la translocation maternelle t(5;14)(q13;q23q32). *Ann Genet (Paris)* 1981;24:176-8.
- Nikolis J, Ivanovic K, Kosanovic M. Tandem duplication of chromosome 14(q24→q32) in male newborn with congenital malformations. *Clin Genet* 1983;23:321-4.
- Pfeiffer RA, Buttinghaus K, Struck H. Partial trisomy 14 following a balanced reciprocal translocation t(14q-;21q+). *Humangenetik* 1973;20:187-9.
- Romain DR, Columbano-Green LM, Smythe RH, et al. Partial trisomy 14q24→qter. *J Med Genet* 1983;20:466-7.
- Sklower S, Jenkins E, Nolin S, et al. Familial distal trisomy 14q. *Am J Hum Genet* 1982;34:145A.
- Trunca C, Opitz JM. Pericentric inversion of chromosome 14 and the risk of partial duplication of 14q (14q31→14qter). *Am J Med Genet* 1977;1:217-28.
- Wyandt HE, Magenis RE, Hecht F. Abnormal chromosomes 14 and 15 in abortions, syndromes, and malignancy. In: Yunis J, ed. *New chromosomal syndromes*. New York: Academic Press, 1977:301-38.

Correspondence and requests for reprints to Dr Daisy M Carr, Cytogenetics Laboratory, Department of Pathology, Charles Drew Medical School, 1621 E 120 St, Los Angeles, California 900059 USA.