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Deletion of chromosome 2 (p11–p13): case report and review

V P Prasher, V H R Krishnan, D J Clarke, C T Maliszewska, J A Corbett

Abstract

The case of a young man with del(2)(p11.2p13) is reported. Accounts of previous cases of deletion of the short arm of chromosome 2 are reviewed. Common features include mental retardation, proportional short stature and weight, dysmorphic facial features (a prominent nose, abnormal ears), and abnormal hands. Growth and developmental delay are present during the postnatal period.

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Structural deletions of the short arm of chromosome 2 alone are rare and only eight cases have previously been reported. Ferguson-Smith *et al*[1] were the first to report a patient with a deletion of the short arm of chromosome 2. This case was also associated with partial duplication of chromosome 5. Later Zackai *et al*[2] reported a case with a monosomy 2p deletion. The description of the phenotype was limited, although further details of the patient were subsequently given by Emanuel *et al*[3] and Neidich *et al*.[4] Other cases of del(2p) have subsequently been reported.

The case of a 32 year old man with karyotype del(2)(p11p13) is reported and two other previous case reports of del(2p), by Fryns *et al*[5] and Duca *et al*,[6] involving the proximal region are reviewed.

Case report

The proband, a 32 year old Caucasian male, is a long term resident of a hospital for the mentally handicapped. He was admitted in 1978 at the age of 19 years. Little is known about his birth because he was taken into care by the local social service department soon after the event. Developmental milestones were delayed; he was unable to walk at the age of $2\frac{1}{2}$ years, made babbling sounds at 2 years, and had poor feeding skills at 3 years. At the age of 5 years his intelligence quotient (IQ) was between 37 and 40 on the Terman-Merrill scale. His early life was spent in different social services' residential settings but because of his mental handicap and behavioural difficulties he was admitted to a hospital for children with mental handicap at the age of 7 years. Reassessment of his intellectual functioning using the Wechsler Intelligence for Children scale gave a verbal IQ score below 45 and a performance score below 44.

During the next six years, with consistent care, gradual improvement occurred in his behaviour. At the age of 13 years he was transferred to a community children's home and there were no major difficulties until the age of 18 years when he began to steal money and other valuable items. He was also verbally abusive and physically aggressive. Neuroleptic medication was of minimal benefit, and he was admitted to the hospital when fire setting became a problem.

Since his admission to the hospital, his antisocial behaviour has been difficult to control and there have been numerous incidents of aggression, theft, and other behavioural problems. Neuroleptic medication along with a behavioural approach has had little long term benefit.

At the age of 32 years his weight is 70 kg (>50th centile), height 180 cm (>50th centile), and head circumference 53 cm (<25th centile). On examination he is a tall, thin man

Department of Psychiatry, Birmingham University, Queen Elizabeth Psychiatric Hospital, Mindelsohn Way, Edgbaston, Birmingham B15 2QZ, UK.
V P Prasher
V H R Krishnan
D J Clarke
C T Maliszewska
J A Corbett

Correspondence to Dr Prasher.

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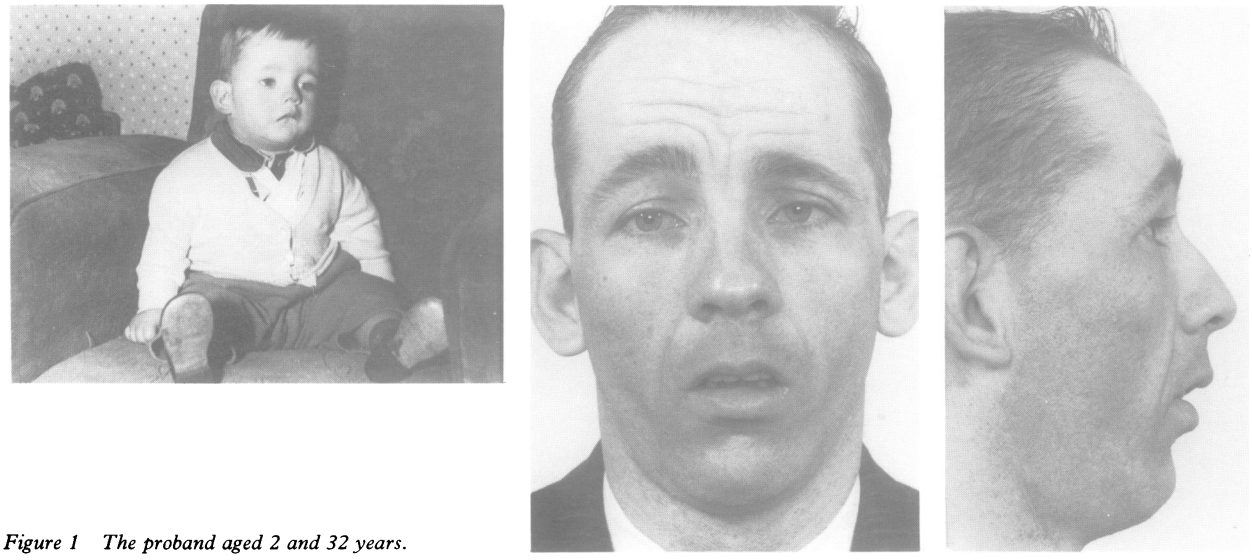


Figure 1 The proband aged 2 and 32 years.

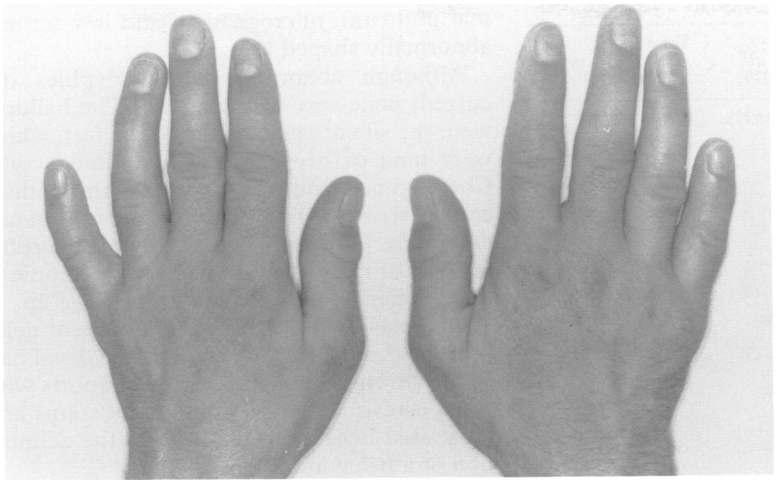
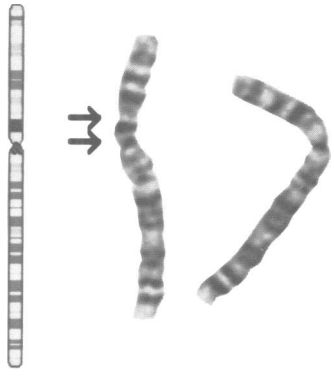


Figure 2 Dorsal view of the hands.

with noticeable facial dysmorphism. He has a narrow, rectangular face, large, protruding, low set ears, minimal blepharophimosis, horizontal palpebral fissures, and a convergent strabismus. He has a pronounced nasal bridge and a broad nose with a well formed philtrum. He has a high arched palate, several absent upper teeth, and micrognathia. His hair is sparse (fig 1).

His hands are normal in length (total hand length 18·5 cm, middle finger length 8 cm) with bilateral short fifth fingers, bilateral fourth finger clinodactyly (fig 2), and finger overlap when he attempts to use the hands. There is partial restriction of movement in all distal interphalangeal joints. Dermatoglyphics on the right are (from the radial side of the hand): whorl, whorl, whorl, radial loop, whorl; on the left (from the radial side of the hand): whorl, whorl, ulnar loop, whorl, radial loop. Three palmar creases are present on both hands.

There are no cardiac, abdominal, or genital abnormalities. His chest is narrow with increased anteroposterior diameter. Neurological evaluation showed reduced visual acuity but otherwise normal cranial nerve function. Power, sensation, and reflexes are normal in all limbs but hypertonia was detected in both legs resulting in a spastic ataxic gait. Full blood count was normal except for a persistent, non-symptomatic, idiopathic thrombocytopenia (platelet counts: 140×10^9/l in 1983, 122×10^9/l in 1985, and 129×10^9/l in 1989; normal range 150 to 400×10^9/l). Biochemical profile and thyroid function tests were normal. Acid phosphatase was normal (the locus for the enzyme acid phosphatase has been assigned to distal 2p). Skull *x* ray was unremarkable but *x* ray of the hands showed a short middle phalanx of both fifth fingers.



46,XY,del(2)(p11.2p13)

Figure 3 Karyotype of the proband.

CYTOGENETIC STUDIES

Cytogenetic studies were performed on stimulated lymphocyte cultures. G banding showed a male karyotype with an interstitial deletion of material in the short arm of one chromosome 2: 46,XY,del(2)(p11.2p13) (fig 3). Therefore the patient is monosomic for part of the short arm of chromosome 2 (band p11.2–p13). A cell line from this patient has not been established.

Discussion

Two cases, by Ferguson-Smith *et al*[1] and by Young *et al*,[7] were excluded from comparison with our case as both had chromosomal abnormalities of other chromosomes as well as the deletion of chromosome 2. The cases reported by Lundbech and Thogerson[8] and Gorski *et al*[9] were excluded from comparison as they had deletions of the short and long arms of chromosome 2. Two further cases, by Munke *et al*[10] and Wilson *et al*,[11] were also excluded as both patients had holoprosencephaly preventing

Table 1 Clinical findings reported in deletions involving proximal 2p.

	Fryns *et al*[5] (1979)	Duca *et al*[6] (1979)	Present case (1992)
Breakpoints, del(2)	(p11p21)	(p13p15)	(p11–p13)
Sex	M	F	M
Birth weight <25th centile	+	+	?
Birth length <25th centile	?	?	?
OFC <25th centile (at birth)	?	?	?
Maternal age at birth	25 y	30 y	?
Maternal karyotype	?	–	?
Paternal age at birth	44 y	29 y	?
Paternal karyotype	–	–	?
Family history	–	–	?
Prenatal history	?	?	?
Feeding difficulties	+	?	+
Failure to thrive	+	?	+
Neonatal muscle tone	Hypo	?	?
Delayed closure of fontanelles	?	+	?
Developmental delay	+	+	+
Stature =<50th centile	+	+	–
=<25th centile	–	+	–
OFC =<50th centile	+	–	+
=<25th centile	+	–	+
Weight =<50th centile	+	+	–
=<25th centile	+	+	–
Mental retardation	+	+	+
Age at report (y)	14	5	32
Age at death	?	?	Alive

+ = feature present. – = feature absent. =< = equal to or less. ? = no information.

Table 2 Reported facial dysmorphism and systemic findings in deletions involving proximal 2p.

	Fryns *et al*[5] (1979)	Duca *et al*[6] (1981)	Present case (1992)
Breakpoints, del(2)	(p11p21)	(p13p15)	(p11–p13)
Dysmorphic face	+	+	+
Abnormal head shape	−	+	+
Forehead			
Prominent	−	−	−
Narrow	+	+	−
Blepharophimosis	−	−	−
Strabismus	+	+	+
Abnormal nasal bridge	−	+	+
Abnormally shaped nose	−	−	+
Abnormal philtrum	−	+	+
Arched palate	−	−	+
Micrognathia	−	−	+
Low set ears	−	+	+
Abnormal ears	−	+	+
Short neck	+	+	−
Sparse hair	−	−	+
Fingers			
Short	−	−	5th
Overlapping	−	−	+
Clinodactyly	−	−	5th
5th finger, single flexion crease	−	−	−
Immobility, Interphalangeal joint	−	−	+
Abnormal dermatoglyphics	−	−	−
Halluces			
Long	+	+	−
Broad	+	+	−
Clinodactyly	+	+	−
Overlapping toes	−	+	−
Vertebral anomalies	+	+	−
Chest abnormalities	+	+	+
Cryptorchidism	+	−	−
Spasticity of legs	+	−	+
Seizures	−	−	Past history

+ = feature present. − = feature absent.

valid behavioural comparison. Comparison of the findings reported by Fryns *et al*[5] and Duca *et al*[6] (table 1) shows no relationship between paternal age, paternal karyotype, family inheritance, or prenatal development and deletion of the proximal part of the short arm of chromosome 2.

A brief review of case reports (tables 1 and 2) shows a variety of features. Developmental delay and mental retardation were present in all cases, with growth retardation present in all cases except in the present reported case. The facial dysmorphism, present in all cases, consisted of an abnormal shape, often rectangular. Prominence of the forehead was observed in our case and a narrow forehead in two cases.[5,6] In general the following abnormalities were found: strabismus, abnormal nasal bridge, abnormally shaped nose, arched palate, abnormal philtrum, micrognathia, and low set and abnormally shaped ears.

Although abnormal dermatoglyphics occurred, none was characteristic. The halluces were the site of abnormality of the feet, which were long or broad or showed clinodactyly. Chest abnormalities occur. Cryptorchidism and spasticity of the legs were reported in one case each. However, none of the dysmorphic features or malformations is pathognomonic of the deletion of the proximal part of the 2p.

The majority of the reported cases of deletions of chromosome 2 involved the distal half of the chromosome. Further case reports with serial assessments of psychological status and associated behaviour may lead to the delineation of a behavioural phenotype.

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