De novo interstitial deletion of 1p (pter→p34.1::p32.3→qter)

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Abstract

We report a case of a 9 month old girl with a de novo interstitial deletion of 1p, karyotype 46,XX, del(1)(pter→p34.1::p32.3→qter). She had dysmorphic features including upward slanting palpebral fissures, a bulbous nose, a long philtrum, low set and malformed ears, a short neck, hypoplastic nails on both index fingers, widened interdigital spaces between the toes, dilated lateral ventricles, right hydronephrosis, a dilated right ureter, mental and motor developmental delay, and generalised hypotonia.

Chromosome abnormalities involving 1p are rare. In particular, partial monosomy for 1p, not associated with a translocation or ring chromosome, has been described to date in only eight patients.¹⁻⁸ We report a case of a girl with multiple anomalies and an interstitial deletion of 1p, which is the smallest of these deletions reported so far.

Case report

The patient, a 9 month old female infant, was born to a 21 year old mother and 31 year old father after an uneventful 40 week pregnancy with a birth weight of 3110 g.

When she was first seen by us at the age of 27 days, she showed the following abnormalities: prolonged jaundice, two strawberry haemangiomas on the parietal area of the scalp, a flat occiput, mildly dilated

lateral ventricles and low density areas in the white matter shown by computed tomography, protuberant orbital areas with epicanthus, short and upward slanting palpebral fissures, a bulbous nose with anteverted nostrils, a long philtrum, a beaked upper lip, a high arched and narrow palate, low set, malformed, and prominent ears, a short neck, widely spaced nipples, hypoplastic nails on both index fingers, long halluces, wide interdigital spaces between the first and second and the fourth and fifth toes,

High resolution GTG banded chromosome 1. Black arrows show breakpoints and white arrow shows a rejoining point.

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Selected clinical features observed in patients with *del(1)(p32.3→pter)*.

Clinical features	Deleted segments		
	Geňčík and Geňčíkova ¹⁰ (pter→p33)	Howard and Porteus ⁸ (p36.1→p34.1)	Present patient (p34.1→p32.3)
Flat occiput	+	—	+
Low set ears	+	+	+
Accessory ears	—	+	—
Slanting palpebral fissures	+	—	+
Small eyes	—	+	+
Bulbous nose/wide nasal alae	—	+	+
High arched palate	—	+	+
Short neck	+	+	+
Hypertrichosis	+	—	—
Cryptorchidism	+	+	Female
Cardiac defects	—	—	—
Developmental retardation	+	+	+

right hydronephrosis, and a dilated right ureter. Her developmental milestones were generally delayed; she had gained head control by 4 months, but could not roll over, sit, or stand until 11 months, and started to walk at the age of 22 months.

Cytogenetic findings and discussion

Cytogenetic analysis with high resolution GTG banding showed the karyotype 46,XX,del(1)(pter→q34.1::p32.3→qter) de novo (figure), and *AluI* banding⁹ indicated that the origin of the de novo deletion was paternal.

One patient has been reported with interstitial deletion⁸ and another with terminal deletion¹⁰ of 1p whose deleted segments showed partial overlap with the deleted region in our patient. The present patient shared some clinical features with these two reported patients, including a flat occiput, low set ears, upward slanting palpebral fissures, short neck and developmental delay (table). At least seven other patients with interstitial deletion^{2,3,5,7} or terminal deletion^{1,4,6} of this chromosome have been reported to date. However, the deleted region in our patient did not show any overlap with the deleted bands of any of these patients.

The size of the deletion in our patient was the smallest of the eight reported deletions of 1p. The small number of reported cases of 1p deletion may

suggest the existence of genes essential for life being located on this chromosome.

An EB virus transformed lymphoblastoid cell line from this patient is available at the Department of Human Genetics, Nagasaki University School of Medicine (No NG-865).

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