

A 46,XX,del(20)(q11.23q13.11) with normal adenosine deaminase (ADA) activity

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Abstract

We present a patient with 46,XX,del(20)(q11.23q13.11). The proband, a female infant, was born after 41-weeks gestation. The birth weight was 3,082 g, length 51.0 cm and head circumference 34.2 cm. Physical findings included: deep-set eyes, anteverted nostrils, a portwine stain on the forehead, low-set ears, bilateral high axial triradius, right hip dislocation, and general muscular hypotonia. There was no history of recurrent infection. Development quotient (DQ) at 7 months after birth was about 60. The patient's G-banded karyotype analyzed on cultured lymphocytes revealed she had an interstitial deletion of 20q (q11.23→q13.11). The level of the patient's adenosine deaminase (ADA), for which the gene locus is mapped on 20q13.11, was 14.6 IU/L (normal range: 6.8-18.2)

Key words : chromosome 20, partial monosomy 20q,
adenosine deaminase (ADA)

Introduction

Structural abnormalities of chromosome 20 are rare. A few cases have been reported with ring 20, a few with complete or partial trisomy (mostly of the short arm), and a very few with partial deletion of the short or the long arm. Deletion of the long arm in particular is extremely rare. To our knowledge, only 2 cases have been reported^{1,2)}. We present an additional case with interstitial deletion of 20q. We also discuss clinical features and the gene locus of adenosine deaminase (ADA), which is mapped on 20q.

Clinical and cytogenetic findings

The female infant proband, the second child of healthy

and unrelated parents, was born after 41-weeks gestation. The birth weight was 3,082 g, length 51.0 cm, and head circumference 34.2 cm. The father was 36 and the mother was 28 years of age at the time of the birth. The elder sister was healthy at 4 years old. The mother had not experienced any spontaneous abortions or still births prior to the proband's birth. Because of poor weight gain, multiple external congenital anomalies, muscular hypotonia, poor feeding and right hip dislocation, she was referred to us at the age of 7 months. At that time she weighed 6,845 g (-2.0 SD), length was 69.1 cm (almost average) and head circumference was 43.2 cm (almost average). Physical findings included the following: deep-set eyes, anteverted nostrils, a portwine stain on the forehead, low-set ears, bilateral high axial triradius, right hip

dislocation, and general muscular hypotonia (figure 1). Her sucking of milk was poor and tube feeding was required. There was no history of recurrent infection. Development quotient (DQ) at 7 months was about 60. The patient's G-banded karyotype analyzed on cultured lymphocytes revealed 46,XX,del(20)(q11.23q13.11). Accordingly she had an interstitial deletion of 20q (q11.23→q13.11) (figure 2). The maternal karyotype was normal; however, the paternal karyotype was unavailable. The level of the patient's adenosine deaminase (ADA), for which the genetic locus is mapped on 20q13.11, was 14.6 IU/L (normal range: 6.8-18.2).

Discussion

Previously reported cases with 20q- and their deleted segments are summarized in figure 3. Since the number of reported cases is small and deleted segments are different, it is natural that clinical features have yet to be established. Shabtai et al.¹⁾, however, from their patient (q13.11→q13.33) and the patient of Fraisse et al. (q13.11→qter)³⁾, concluded that it seemed possible to delineate a 20q- syndrome when the deletion included the q13.12→q13.32 region, the major features being retardation, severe malformations of the limbs, short neck, flat occiput, and mild facial dysmorphism. These cases can be called a distal 20q- syndrome. Another case with a small terminal deletion of 20q (q13.3→qter) showed intrauterine growth retardation, dolicocephaly, hypotonia, cleft palate, congenital heart defects, a subependymal cyst, and hypospadias⁴⁾. However, this case had partial trisomy 16q (q22.1→qter) simultaneously; thus seemingly having no distinct dysmorphic syndrome. On the other hand, the patient of Petersen et al.²⁾ had the same range of deletion as our patient (q11.23→q13.11). Their case showed growth and developmental retardation, mild facial dysmorphism including low set ears, broad nasal bridge, and macrostomia, heart murmur, a convergent strabismus, febrile seizures, and

retarded bone age. From this case and ours, the proximal 20q deletion may be characterized by growth and developmental retardation, with a peculiar facial appearance that includes low set ears, broad nasal bridge, deep set eyes, and macrostomia.

The gene locus of human adenosine deaminase (ADA) has been assigned to the q12-q13.11 region of chromosome 20⁵⁾. Both the patient of Fraisse et al.³⁾ and that of Petersen et al.²⁾ had ADA deficiency, which led Petersen et al.²⁾ to conclude the ADA gene must be located at 20q13.11. Because our patient had a normal ADA level, the gene locus of the ADA must be mapped at the distal portion of within the band 20q13.11 near band 20q13.12.

References

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Fig.1. Proband at 7 months of age (published with permission of the child's parents)

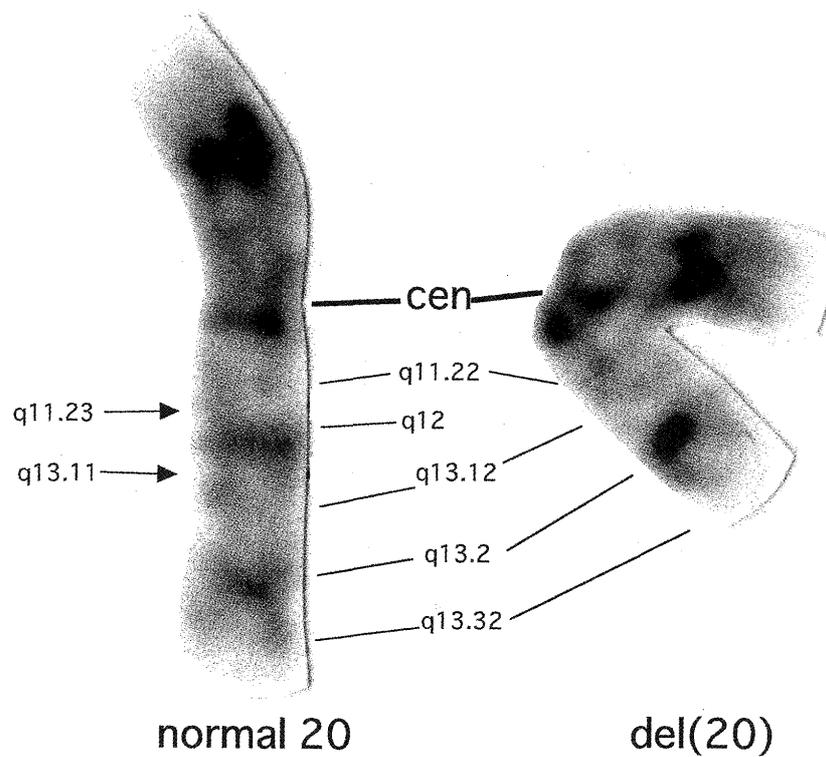


Fig.2. Partial karyotype of the proband

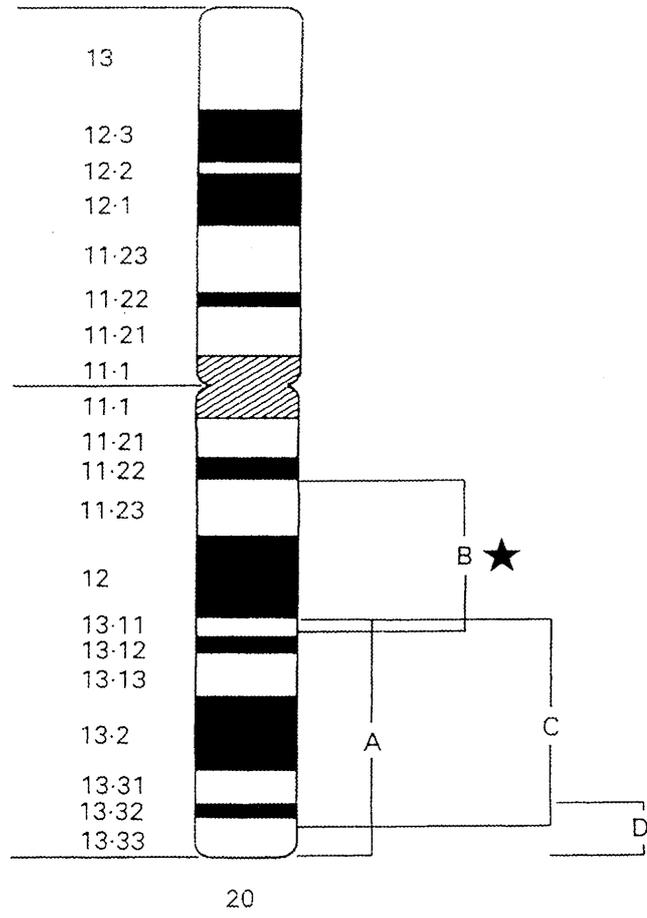


Fig.3. Schematic representation of the 20q deletions reported in different cases; A Fraisse et al.³⁾, B Petersen et al.²⁾, C Shabtai et al.¹⁾, D Chen et al.⁴⁾, ★ present case

正常adenosine deaminase (ADA)活性を伴う 46,XX,del(20)(q11.23q13.11)の1例

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要旨

46,XX,del(20)(q11.23q13.11)の1例を報告する。症例は在胎41週1日で出生した。出生体重は3,082 g、身長51.0 cm、頭囲34.2 cmであった。眼球陥凹、前向き鼻孔、前額部のポートワインマーク、耳介下方付着、両側軸三叉線高位、右股関節脱臼、全身の筋緊張低下などの異常がみられた。反復性感染の既往はなかった。生後7カ月のときの発達指数は約60であった。末梢血リンパ球を用いたGバンドによる染色体検査では、患児は20q11.23→q13.11の腕内欠失を有していることがわかった。20q13.11に遺伝子座があるといわれるadenosine deaminase (ADA)値は14.6 IU/L (正常値6.8-18.2)であった。

キーワード：20番染色体長腕部分モノソミー、adenosine deaminase (ADA)