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SHORT CLINICAL REPORT: A NEW CASE WITH DE NOVO PARTIAL 9_p MONOSOMY

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A female patient is described with a karyotype 46,XX,del (9) (p22) showing characteristic dysmorphic phenotype: trigonocephaly, prominent forehead, long philtrum, small mouth, high arched palate, low set ears, short neck, widely spaced nipples, long fingers and toes, omphalocele.

The first Hungarian case of 9p monosomy syndrome is reported here.

INTRODUCTION

The 9p monosomy syndrome was first delineated by Alfi in 1973 using two cases /2/ and was further described on the basis of six cases /3/. Until 1986, fewer than 60 cases were reported /15/. The clinical features of the syndrome are consistent and characteristic /3, 8, 22, 24/. The most common manifestations are summarized in Table I.

Life expectancy does not seem significantly diminished /8, 19/.

The 9p monosomy occurs more often in female than male patients /8, 24/.

In most of the cases the aberration is de novo /1, 2, 3, 9, 11, 15, 18, 23/, but it may be inherited /2, 12, 20/ and/or combined with other aberrant chromosome /7, 10, 12, 17/. One case was reported having ring (9)/del (9p) mosaicism /15/.

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TABLE I

The main clinical features in patients with 9p- deletion syndrome

Clinical findings from previous reports	Present case
Moderate mental and developmental retardation	+
Hypertonia	-
Trigonocephaly	+
Prominent forehead	+
Flat occiput	+
Up-slanting palpebral fissures	+
Epichantal folds	+
Ocular hypertelorism	+
Exophthalmos	+
Flat nasal bridge	+
Anteverted nostrils	+
Long philtrum	+
Small mouth	+
Micrognathia/retrognathia	+
High-arched palate	+
Flat abnormal auricles	+
Low-set ears	+
Short broad neck	+
Widely spaced nipples	+
Cardiovascular malformations	-
Square hyperconvex nails	-
Long fingers and/or toes	+
Omphalocele	+

CASE REPORT

The proband, a girl born on 25th of June 1989, is the first child of healthy unrelated parents. Mother was 20 years old, and the father was 24 at the time of her birth. The pregnancy and delivery were uneventful. The birth weight was 4350 g. Apgar score was 8/9. Maternal serum AFP level was higher than normal during the 16th week of pregnancy, but there was not shown any abnormalities by ultrasound. Omphalocele was seen immediately after birth, therefore she was sent for operation.

Cytogenetic examination was performed because of her multiple congenital abnormality including craniofacial dysmorphism (Fig. 1 and Table I). Muscle tone was normal. There was no hyperreflexia.

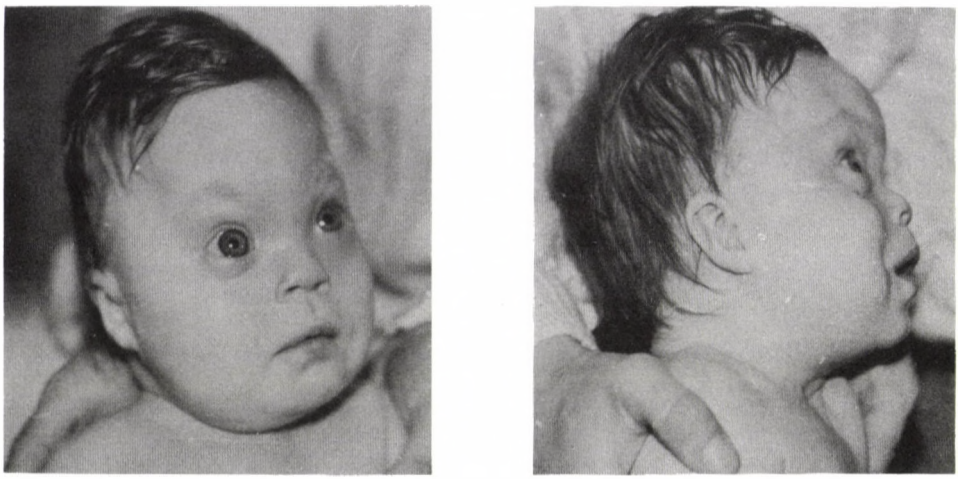

Fig. 1. Frontal /a/ and lateral /b/ views of the patient aged 4 months.

Psychological test /13, 14/ was performed at the age of 9 months. According to this test she was delayed mentally and developmentally. She was functioning at six months level. However she is amiable, affectionate and sociable.

Cytogenetics

Chromosome investigations of the patient and her parents were performed on peripheral blood leukocytes using standard phytohemagglutinin-stimulated whole-blood cultures. Cytogenetic analysis of the proband's cells demonstrated a terminal deletion of the short arm of chromosome 9 at band p 22, identified by G- and C-banding (Fig. 2). The karyotype was 46,XX,del (9) (p22). Parents' chromosomes were normal.

DISCUSSION

Clinical features of 9_p- syndrome are consistent and characteristic. However, unusual clinical features can be found in some cases, i.e. advanced osseus maturation, marked congenital vertebral anomalies /21/ and redundant posterior neck skin, absence of exophthalmos /15/. This may be related to the fact, that patients have other chromosome aberration beside 9p deletion /10, 15/. Our case shows the most characteristic phenotype of the syndrome (Table I) and she does not have any distinctive features.

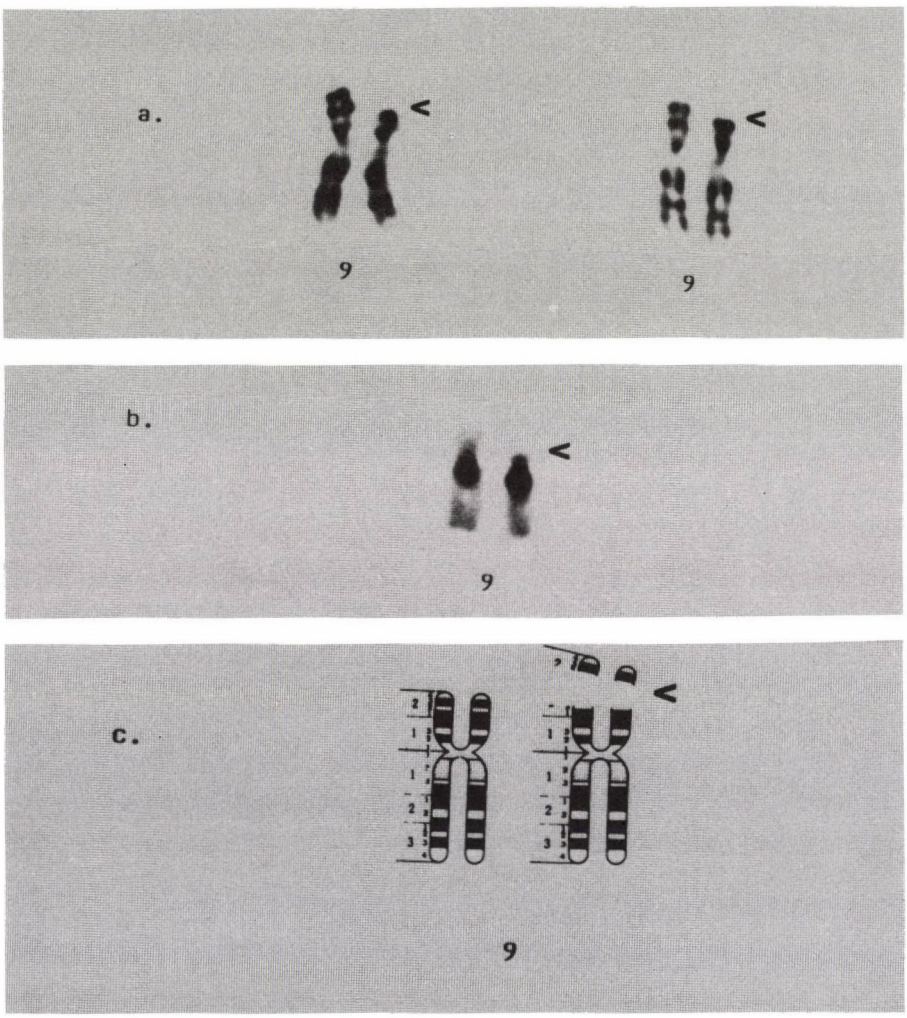

Fig. 2. Three partial karyotypes of the patient using G-banding /a/ and C-banding /b/ techniques, and the idiogram of the aberration /c/. Arrowheads show the breakpoint on the short arm of chromosome 9 at band p22.

Patients' apparently long fingers due to relative shortness of metacarpals /23/ were found in some patients /1/.

The GALT (galactose-1-phosphate uridyl transferase) activity was studied in two patients with 9p deletion. Findings

suggested that locus of the GALT gene is on the short arm of chromosome 9 in band 9p21 /5/.

9p deletion was found in patients with acute lymphoblastic leukemia (ALL) as well /4, 6, 16/.

Coordinated studies will be necessary to clarify the importance of chromosomal material at 9p21-22.

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REFERENCES

1. Al-Awadi SA, Farag TI, Satyanath S, et al: Letter to the editor: Metacarpophalangeal pattern profile analysis in partial 9p monosomy. Am J Med Genet 29: 217, 1988

2. Alfi OS, Donnell GN, Crandall BF, et al: Deletion of the short arm of chromosome 9 (46,9p-): A new deletion syndrome. Ann Genet 16: 17, 1973

3. Alfi OS, Donnell PWA, Derencsenyi A: The 9p- syndrome. Ann Genet 19: 11, 1976

4. Bigner SH, Friedman HS, Kinney TR, et al: 9p- in a girl with acute leukemia and sickle cell disease. Cancer Genet Cytogenet 21: 267, 1986

5. Bricarelli FD, Magnani M, Arslanian A, et al: Expression of GALT in two unrelated 9p-patients. Evidence for assignment of the GALT locus to the 9p21 band. Hum Genet 59: 112, 1981

6. Chilcote RR, Brown E, Rowly JD: Lymphocytic leukemia with lymphomatous features associated with abnormalities of the short arm of chromosome 9. N Engl J Med 313: 286, 1985

7. Daniel A, Ekblom L, Phillips S, et al: NOR activity and centromere suppression related in a de novo fusion t dic (9;13) (p22;p13) chromosome in a child with del (9p) syndrome. Am J Med Genet 22: 577, 1985

8. De Grouchy J, Turleau C: 9p monosomy. In: Clinical Atlas of Human Chromosomes, ed John Wiley and Sons Inc., New York-Chicester-Bisbane-Toronto 1977, p. 87

9. Deroover J, Fryns JP, Parloir C, et al: Clinical case reports. Partial monosomy of the short arm of chromosome 9. A distinct clinical entity. Hum Genet 44: 195, 1978

10. Eden MS, Thelin JW, Michalski K, et al: Partial trisomy 6p and partial monosomy 9p from a de novo translocation 46, XY,-9,+der (9) t (6:9) (p11:p24). Clin Genet 28: 375, 1985

11. Funderburk SJ, Spakers RS, Klisak I: The 9p- syndrome. J Med Genet 16: 75, 1979

12. Game K, Friedman JM, Kalousek DK: Mild phenotypic abnormalities in combined del 9p2 and dup 3p2. Am J Med Genet 35: 370, 1990

13. Hegedüs TA, Szondy M, Popper P: A csecsemő- és kisgyermekkori fejlődés magyar standardja I. Magy Pszichol Szle 32: 489, 1975

14. Hegedüs TA, Szondy M, Popper P: A csecsemő- és kisgyermekkori fejlődés magyar standardja II. Magy Pszichol Szle 32: 581, 1975

15. Leung AKC, Rudd NL: A case of ring (9)/del (9p) mosaicism associated with gastroesophageal reflux. Am J Med Genet 29: 43, 1988

16. Maseki N, Hayashi Y, Kaneko Y, et al: Partial loss of the short arm of cromosome 9 in leukemia and lymphoma. Cancer Genet Cytogenet 22: 89, 1986

17. Mattei JF, Mattei MG, Balestrazzi P, et al: Letters to the editors. Familial paracentric incersion of chromosome 9, inv (9) (p22q32) with recurrent duplication-deletion. Clin Genet 24: 220, 1983

18. Mulachy MT: Another case of 9p- syndrome. Ann Genet 21: 47, 1978

19. Nielsen J, Homma A, Christiansen F, et al: The deletion 9p syndrome. A 61-year-old man with deletion of short arm 9. Clin Genet 12: 80, 1977

20. Orye E, Verhaaren H, Vandenbogaert AM: The 9p- deletion syndrome. Report of a patient with a 46,XX,9p- constitution due to a paternal t(9p-;15q+) translocation. Clin Genet 8: 349, 1975

21. Rutten FJ, Hustinx TWJ, Dunktillemans AAW, et al: A case of partial 9p monosomy with some unusual clinical features. Ann Genet 21: 51, 1978

22. Tiwary CHM, Surana RB: The 9p- with excess of digital arches. Pediatr Res 16:2:196/A, 1982

23. Young RS, Bader P, Palmer CG, et al: Brief clinical report: Two children with de novo del 9p. Am J Med Genet 14: 751, 1983

24. Young RS, Reed T, Hodes ME, et al: The dermatoglyphic and clinical features of the 9p trisomy and partial monosomy syndromes. Hum Genet 62: 31, 1982

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