Trisomy 7p Resulting From 7p15;9p24 Translocation: Report of a New Case and Review of Associated Medical Complications

Chahira Kozma,1* Bassem R. Haddad,2 and Jeanne M. Meck2

¹Child Development Center/Department of Pediatrics, Georgetown University Medical Center, Washington, D.C. ²Departments of Oncology and Obstetrics and Gynecology, Georgetown University Medical Center, Washington, D.C.

The authors report on a young girl with generalized developmental deficits originally thought to be caused by an unusual reaction to DPT vaccination. At the age of 4½ years, chromosome analysis showed that the terminus of the short arm of chromosome 9 had extra material believed to originate from 7p terminus, thus she was considered to be trisomic for a segment of 7p and monosomic for a small portion of 9p [46,XX,der (9), t(7;9)(p15;p24)]. Ten years later, molecular cytogenetic testing using fluorescence in situ hybridization (FISH) confirmed that the extra chromosomal material represented partial trisomy 7p. The proposita had a high and large forehead, hypertelorism, and broad nasal bridge, findings seen in most individuals with trisomy 7p. Longterm follow-up showed the presence of hypothyroidism, obesity, and cerebral palsy. A review of all published cases of trisomy 7p with focus on associated complications suggests a well-defined pattern of abnormalities characterized by musculoskeletal, cardiovascular, neurological, genital, and ocular abnormalities in decreasing frequency. At least one-third of affected individuals died in infancy and close to half had severe mental retardation. FISH was essential in the confirmation of the cytogenetic abnormality and further delineation of the chromosomal disorder. Am. J. Med. Genet. 91:286-290, 2000. © 2000 Wiley-Liss, Inc.

KEY WORDS: trisomy 7p; chromosome 7; chromosome 7 duplication; macrocephaly; hypothyroidism; infantile spasms; FISH; wide fontanelle; wide sutures

INTRODUCTION

Trisomy 7p causes psychomotor delay, macrocephaly, large and high forehead, persistently open anterior fontanelle, hypertelorism, and apparently lowset ears in addition to abnormal palmar creases and cardiovascular anomalies. Over 37 cases of confirmed partial trisomy 7p have been described, most resulting from malsegregation of familial translocations [Lurie et al., 1995]. However, the natural history of 7p trisomy is not well defined, since most published cases describe affected individuals in early infancy and childhood with minimal data on natural history.

We describe a 14½-year-old girl with partial trisomy 7p and provide long term follow-up with documentation of associated medical complications including hypothyroidism, Duane syndrome, foot deformity, cerebral palsy, and obesity. We reviewed all published cases of trisomy 7p and associated medical complications in an effort to provide medical characterization of the syndrome.

CLINICAL REPORT

The proposita was evaluated at the age of 4½ years for developmental deficits. Mother and father were 30 and 24 years old, respectively, at the time of conception. The pregnancy was unremarkable. Birth weight was 2.8 kg (10th–25th centile) and length was 50 cm (50th centile). Postnatally, she experienced feeding difficulties and inadequate weight gain. Anterior fontanelle and sutures were reported to be very wide in infancy. According to the parents, following the administration of the second dose of diphtheriatetanus-pertussis (DPT) vaccine at the age of 4 months, she developed irritability, excessive crying, an increase in the frequency of deep sleep, and "abdominal symp-

^{*}Correspondence to: Chahira Kozma, M.D., Child Development Center, Georgetown University Medical Center, 3307 M Street, N.W. Suite #401, Washington, D.C. 20007-3935. E-mail: kozmac@gunet.georgetown.edu

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toms." She was evaluated in a hospital where the diagnosis of infantile spasms was made based on history and an abnormal electroencephalogram (EEG). Computerized tomography (CT) scan of the head demonstrated large ventricles. Seizure activities ceased following treatment with corticosteroids for 9 weeks. An eye evaluation documented Duane anomaly in the left eye. Hearing was normal. She had recurrent upper respiratory tract infections and constipation. When she was several months old she was enrolled in an early infant intervention program.

The parents indicated that psychomotor development was normal before the onset of infantile spasms. She smiled at the age of 2 months and rolled over at 4 months. Delayed head control was attributed to macrocephaly. She sat at 9 months, crawled at 15 months, and walked independently at 21 months. Her expressive speech was very limited. A developmental evaluation at the age of 20 months indicated a functional level between 9 to 10 months. The family history was unremarkable; the older sister was reported to be normal.

On physical examination at the age of 4½ years, her occipitofrontal circumference (OFC) was 54.2 cm (>98th centile), height 101 cm (25th centile), and weight 17.6 kg (50th centile). She had partial alopecia, prominent forehead and glabella, and hypertelorism. The outer canthal distance was 9.4 cm (>97th centile) and inner canthal distance 3.8 cm (>97th centile). Ears measured 5.8 cm (25th centile). Her palate was normal and she had inverted nipples (Fig. 1). Thumbs and fingers were broad, and her feet were flat. Each hand measured 11.5 cm (10th centile), and each foot measured 15 cm (10th centile). The proposita displayed generalized hypotonia, increased deep tendon reflexes, and four to five bouts of clonus bilaterally. A diagnostic workup (kidneys sonogram, thyroid function tests,



Fig. 1. The proposita at the age of 4 years. Note the prominent forehead and hypertelorism.

metabolic screening of the urine, and blood chemistry) yielded normal results. Hearing was normal. Repeated EEG showed a mild diffuse slowing of background activity without epileptiform signs. On the Peabody Fine Motor Scale, she received an overall age equivalent of 21 months. She had poor postural control, weak antigravity ability, weak concentration, and poor motor planning. Cognitive testing on the Bayley Scales of Infant Development (Mental Scale) yielded an overall age score of 19.9 months and a ratio IQ was calculated to be 38 placing her within the moderate range of mental retardation. Weaknesses were demonstrated in expressive language, fine motor, and imitative skills. On the Vineland Adaptive Behavior Scale, adaptive functioning was at the 18 month age equivalent, placing her in the moderate range of developmental deficits. Receptive skills scattered from the 14 to 20 month level, and expressive skills from the 7 to 11 month level. She did not demonstrate a functional expressive communication system. Behavioral problems included hair pulling and self-stimulatory and disruptive behaviors.

The proposita was next seen 10 years later. During the interval she had developed hypothyroidism treated with hormone replacement; episodes of depression managed with antidepressant medications; recurrent upper respiratory tract infections; asthma; and obesity. Partial upper airway obstruction responded to tonsillectomy. Repeat kidney sonogram showed normal results, and an EEG showed mild diffuse slowing of electrical activity. Menarche began at age 14 years. However, menses were irregular. She had most of her self-help skills, but needed assistance with showering and bathing. On repeated examination, OFC was 60 cm (>98th centile), height 158 cm (50-75th centile), and weight 100 kg (=98th centile). She had multiple striae on her chest, breasts, deltoid, and thighs and minimal axillary and pubic hair. Breasts were at Tanner Stage IV in development. She had a prominent forehead, malar flattening, inner canthal distance of 3.8 cm (>98th centile), outer canthal distance of 10.5 cm (>98th centile), palpebral fissures 3.4 cm (>98th centile), downslanting palpebral fissures, high arched palate, and malocclusion type II. Ears were posteriorly angulated with an abnormal crease on the right ear lobe; each measured 6.8 cm (75-97th centile). She had no murmurs. She had hyperextensible distal joints and equinovarus deformity of the feet more severe on right than on left. Each hand measured 18.5 cm (75–97th centile) and each foot 24.5 cm (50th-75th centile). Examination of dermatoglyphics showed eight fingers with whorls, whereas the right middle finger and first finger on the left had ulnar/radial loops. Her speech was dysarthric, and she showed oral motor apraxia. She had deficient lateral gaze OD and hyperadduction OS. She had moderate generalized hypotonia with at least 4/5 power. Deep tendon reflexes were brisk. She had a spastic gait and drop foot.

CYTOGENETIC FINDINGS

The initial cytogenetic study prepared from a peripheral blood sample showed extra material on distal 9p

that appeared to consist of a portion of distal 7p (7p15→pter). The terminal band of 9p (9p24) was absent. Thus, the proposita was thought to have partial trisomy of 7p and partial monosomy of 9p. Parents had normal chromosomes. Recently, fluorescence in situ hybridization (FISH) studies were performed in order to confirm our impressions from G-banding. Whole chromosome paints for chromosomes 7 (digoxigeninlabeled) and 9 (biotin-labeled) were hybridized to the patient's metaphases and detected according to the manufacturer's instructions (Oncor, Gaithersburg, MD). Five metaphases were examined. The FISH study unambiguously identified the extra material on 9p as being derived from chromosome 7. Both chromosomes 7 showed normal hybridization signals, and there was no evidence that distal 9p was present on either chromosome 7. These results demonstrate the presence of an unbalanced translocation resulting in partial trisomy 7p and partial monosomy 9p (Fig. 2A-D). The karyotype designation for this unbalanced translocation was as follows: 46, XX, der(9)t(7;9)(p15;p24).ish der (9)t(7;9)(wcp7+,wcp9+).

DISCUSSION

As an increasing number of cases of trisomy 7p are identified with appropriate molecular cytogenetic techniques, a specific phenotype is emerging. A medical profile can be generated with important implications for management. Prenatal and postnatal counseling and provision of anticipatory guidance to at-risk families is contingent on an understanding of the full clinical picture of trisomy 7p.

Thirty-seven cases are published. Table I summarizes the manifestations seen in trisomy 7p patients. The most common medical complications were those of the musculoskeletal system that could be related to abnormal muscle tone in intrauterine life as well as in postnatal life. Indeed, a third of affected individuals

displayed hypotonia or decreased muscle tone. The most common abnormalities consisted of talipes deformities, flexion deformity of fingers, and dislocation of hips with occasional knee pathology. Malformations were infrequent and consisted of agenesis of sacrum [Odell et al., 1987] and hypoplastic pelvis [Moore et al., 1982].

Cardiovascular abnormalities were confirmed in 16 patients (43%), with the most common malformations being atrial and ventricular septal defects and patent ductus arteriosus. It has been hypothesized that more than one segment on 7p may be involved in normal cardiac development [Lurie et al., 1995]. Echocardiogram of the heart should be obtained in all cases of trisomy 7p.

Central nervous system abnormalities were observed in 13 cases (35%). At least five patients had severe CNS abnormalities in fore- and hindbrain development, and, in four of them, the duplicated segment included the proximal segment of 7p(p11p12). It has been postulated that duplications of 7p11p12 may cause defects in fore- and hindbrain development [Lurie et al., 1995]. Several genes that map to 7p are linked to central nervous system (CNS) abnormalities. The gene (CCM2) which appears to be involved in the cerebral cavernous malformations maps to 7p15-13. CCM2 is a Mendelian model of stroke, characterized by focal abnormalities in small intracranial blood vessels leading to hemorrhage and consequent strokes [Craig et al., 1998]. A gene controlling the development of craniosynostosis type 1 (CRS) maps to 7p21.3-p21.2. Recently, mutations in the TWIST gene (ACS3, SCS), which maps to 7p21, were found to result in Saethre-Chotzen syndrome [Johnson et al., 1998]. To date, the genes identified in craniosynostotic syndromes either regulate transcription or are required for signal transduction and play a central role in the development of the calvarial sutures [Muller et al., 1997]. Magnetic resonance imaging (MRI) of the brain is recommended in cases of trisomy

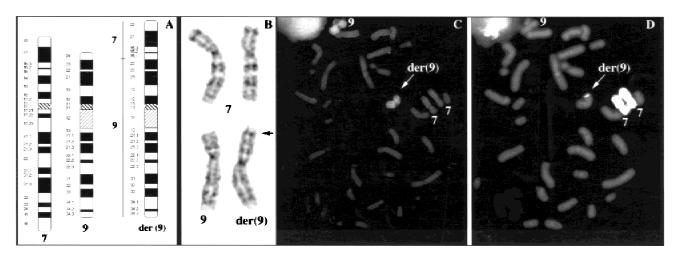


Fig. 2. A: Idiograms of chromosomes 7, 9 and der(9). B: GTG banded partial karyotype of the patient showing chromosomes 7, 9 and der (9), t(7;9)(p15;p24). Arrow indicates the chromosome 7 material translocated to 9p. C: FISH analysis using chromosome 9 painting probe on lymphocyte metaphase spread prepared from the proband. Uniform hybridization on the normal 9 and der(9) is seen, with the exception of the 9qh region on both chromosomes and the distal short arm of der(9). No other hybridization signal is detected on any other chromosome, particularly 7. D: The same metaphase spread shown in C is shown hybridized with chromosome 7 painting probe. Uniform hybridization signal is detected on both chromosomes 7. Chromosome 7 material is also detected on the p arm of the der(9) chromosome, confirming the G-band diagnosis

TABLE I. Trisomy 7 Manifestations

| | Duplicated segment | | | | | | | | | |
|----------------------------------|---------------------|---------------------|---------------------|---------------------|----------------------|-------------------------|----------------------|--|--|----|
| | 7p11-pter $(n = 7)$ | 7p12-pter $(n = 1)$ | 7p13-pter $(n = 4)$ | 7p14-pter $(n = 3)$ | 7p14.2-p21 $(n = 1)$ | 7p15.1-p21.3 (n = 2) | 7p15-pter $(n = 13)$ | $\begin{array}{c} 7p21\text{-pter} \\ (n = 6) \end{array}$ | $\begin{array}{c} Total \\ (n = 37)^a \end{array}$ | % |
| Central nervous system | | | | | | | | | | |
| Hydrocephalus | 2/7 | 0/1 | 0/4 | 0/3 | 0/1 | 1/2 | 3/13 | 1/6 | 7/37 | 19 |
| Other anomalies | 4/7 | 1/1 | 0/4 | 0/3 | 1/1 | 0/2 | 6/13 | 2/6 | 13/37 | 35 |
| Hypotonia | 3/7 | 0/1 | 1/4 | 0/3 | 0/1 | 0/2 | 5/13 | 2/6 | 11/37 | 33 |
| Organ malformations | | | | | | | | | | |
| Ōcular | 1/7 | 1/1 | 1/4 | 1/3 | 1/1 | 0/2 | 4/13 | 2/6 | 11/37 | 30 |
| Oral cleft | 0/7 | 1/1 | 2/4 | 0/3 | 0/1 | 0/2 | 1/13 | 1/6 | 5/37 | 13 |
| Choanal atresia | 1/7 | 0/1 | 1/4 | 2/3 | 0/1 | 0/2 | 2/13 | 0/6 | 6/37 | 16 |
| Cardiac | 5/7 | 1/1 | 3/4 | 1/3 | 0/1 | 0/2 | 4/13 | 2/6 | 16/37 | 43 |
| Renal | 0/7 | 1/1 | 0/4 | 0/3 | 0/1 | 0/2 | 1/13 | 1/6 | 3/37 | 8 |
| Genital | 5/7 | 1/1 | 1/4 | 0/3 | 1/1 | 0/2 | 4/13 | 2/6 | 14/37 | 39 |
| Gastrointestinal | 3/7 | 1/1 | 0/4 | 0/3 | 0/1 | 0/2 | 1/13 | 0/6 | 5/37 | 13 |
| Musculoskeletal system | | | | | | | | | | |
| Foot deformity | 5/7 | 1/1 | 2/4 | 1/3 | 0/1 | 0/2 | 5/13 | 4/6 | 18/37 | 49 |
| Other abnormality | 5/7 | 1/1 | 2/4 | 1/3 | 1/1 | 0/2 | 4/13 | 4/6 | 18/37 | 49 |
| Evolution | | | | | | | | | | |
| Feeding difficulty | 2/7 | 0/1 | 1/4 | 0/3 | 0/1 | 0/2 | 3/13 | 1/6 | 7/37 | 19 |
| Recurrent URTI ^b | 0/7 | 0/1 | 1/4 | 0/3 | 0/1 | 1/2 | 2/13 | 0/6 | 4/37 | 11 |
| Growth retardation | 4/7 | 0/1 | 1/4 | 1/3 | 1/1 | 0/2 | 4/13 | 2/6 | 13/37 | 35 |
| Mild to moderate MR ^b | 2/7 | 0/1 | 1/4 | 0/3 | 1/1 | 1/2 | 1/13 | 1/6 | 7/37 | 16 |
| Severe MR | 4/7 | 0/1 | 2/4 | 0/3 | 0/1 | 1/2 | 6/13 | 3/6 | 16/37 | 43 |
| Early death | 4/7 | 0/1 | 2/4 | 1/3 | 0/1 | 0/2 | 2/13 | 2/6 | 11/37 | 30 |

^aThe group was composed of 34 live births and 3 fetuses.

7p for thorough evaluations of any associated CNS abnormalities.

Genital abnormalities were noted in 12 cases (39%) and consisted mostly of genital hypoplasia. It appears that 7p includes genes that are necessary for normal genital development. HOX genes, which specify developmental boundaries and determine cell fate during morphogenesis have been linked to abnormal genital development. Recently, the hand-foot-genital syndrome was shown to result from a mutation in a HOX gene (HOXA13), which maps to 7p15-p14.2. The syndrome affects the digital structures and can involve uterine anomalies in females and hypospadias of variable severity in males [Innis, 1997; Mortlock and Innis, 1997].

Eye abnormalities were seen in 11 cases (30%) and consisted mostly of mild microphthalmia and ptosis. Duane syndrome was documented for the first time in our patient. Recent data suggests that mutations in the BPES2 gene, which maps to 7p21-p13, cause blepharophimosis, epicanthus inversus, and ptosis [De Baere et al., 1999].

Other systemic abnormalities included choanal atresia (13%), gastrointestinal malformations (13%), oral cleft (13%), and renal abnormalities (8%).

The mortality associated with trisomy 7p is not insignificant, and death in early infancy is seen in close to one-third of cases. All of those who died had congenital malformations of the cardiovascular system. Follow-up data on surviving patients showed severe mental retardation in approximately 40% of cases and mild to moderate mental retardation in 17% of affected individuals.

Although our patient is monosomic for 9p24, she did not have any of the manifestations of 9p deletion. These include trigonocephalic shape of head, an upward slant of the palpebral fissures, a long philtrum, a short nose, and a webbed neck [Teebi et al., 1993].

However she did have obesity, which was seen in one of the cases of 9p monosomy [Serra et al., 1997]. Her predominant facial changes were consistent with trisomy 7p — mostly the large fontanelle in early childhood, prominent forehead, and hypertelorism. Significant enlargement of the fontanelle is the most characteristic finding of most patients with trisomy 7p. It has been postulated that duplications of the various segments of 7p may hamper cranial vault ossification. In addition to obesity and depression, a new finding in our patient is hypothyroidism. Although caution must be exercised not to generalize from one case, we recommend periodic monitoring of thyroid function in individuals with trisomy 7p.

FISH was essential in the certain identification of partial trisomy 7p in the proposita and has permitted us to offer more accurate clinical information on this syndrome to the parents. In addition, the long-term follow-up allowed us to contribute toward a better understanding of the natural history of this syndrome.

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^bURTI: upper respiratory tract infections, MR: mental retardation.

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