Online journal ISSN - 1676-5680
GMR Genetics and Molecular Research
EVOLUTION AND TECHNOLOGY

Patau syndrome with a long survival. A case report

A.C. Duarte, A.I.C. Menezes, E.S. Devens, J.M. Roth, G.L. Garcias and M.G. Martino-Roth

Serviço de Aconselhamento Genético do Departamento de Zoologia e Genética, Instituto de Biologia, Universidade Federal de Pelotas, RS, Brasil
Corresponding author: M.G. Martino-Roth
E-mail: martino@ufpel.tche.br/gmartino@brturbo.com

Genet. Mol. Res. 3 (2): 288-292 (2004)
Received August 6, 2003
Accepted May 7, 2004
Published June 21, 2004

ABSTRACT. Trisomy 13 is a clinically severe entity; 85% of the patients do not survive beyond one year, and most children die before completing six months of age. We report a female child, 28 months old, white, the fourth child of a non-consanguineous couple, who presented trisomy 13. The child was born at term, from a vaginal delivery, weighing 2600 g. At birth, she was cyanotic, icteric, spastic, and cried weakly. The initial clinical examination detected polydactyly in the left hand, congenital clubfoot and convex soles, ocular hypertelorism, a low nasal bridge, numerous hemangiomas distributed throughout the body, cardiomegaly, and perimembranous inter-ventricular communication. There was no cleft lip or palate. On physical examination at 18 months old, the child weighed 6,900 g, had a cephalic perimeter of 41 cm, a thoracic perimeter of 43 cm and was 76 cm tall. At 28 months, she weighed 10,760 g and was 88.5 cm tall. Neuropsychomotor development retardation was evident from birth and, according to the psychologist and the social assistant of APAE (Handicapped Parents and Friends Association) in Canguçu, Rio Grande do Sul, there was a noticeable improvement after physiotherapy and recreational sessions.

Key words: Patau syndrome, Trisomy 13

INTRODUCTION

Trisomy 13 or D_1 trisomy is a clinically severe condition first described by Patau et al. (1960). Eighty-two percent of the patients die within one month (Best, 2002). Eighty-five percent do not survive beyond one year of life and most die before completing six months (Wideremann et al., 1980). There are only five reports of patients with trisomy 13 who have survived past the first decade (Singh, 1990; Tunca et al., 2001). The sex ratio at birth is skewed toward females, presumably because of decreased survival among males, with continued skewing of the ratio further toward females as these children age (Best, 2002).

The frequency of this syndrome is 1:5000 live births. It is the third most frequent trisomy among live births (Jones, 1998). Its importance is due to its association with numerous malformations of the central nervous system, the cardiac and circulatory system, and the urogenital system, in addition to a limited survival rate. Central nervous system malformations are important predicting factors of the survival of a child who bears this syndrome (Goel and Rathore, 2000). The presence of semi-lobar or lobar holoprosencephaly in the bearer may cause early death (SOFT Volunteers, 2002). We describe the case of a female patient with trisomy 13 (47, XX + 13) who survived longer than usual.

CASE REPORT

The patient was female, 28 months of age, white, the fourth child of a non-consanguineous couple. Antenatal care was started at the fourth month of pregnancy, and the mother perceived the first fetal movements during the fifth month. The mother was 36 years old when she became pregnant. She reported the use of oral contraceptive pills during conception, and anemia and urinary tract infection were detected and treated during antenatal care. The child was born at term, from a vaginal delivery, and weighed 2,600 g. At birth, she was cyanotic, icteric, spastic, and cried weakly. The initial clinical examination detected polydactyly in the left hand, congenital clubfoot and convex foot soles. She also presented ocular hypertelorism, a low nasal bridge, numerous hemangiomas distributed throughout the body, cardiomegaly, and perimembranous inter-ventricular communication confirmed by electrocardiogram. There was no cleft lip or palate.

Polydactyly was corrected on the child's third day of life. Jaundice was managed with phototherapy and the left foot was immobilized. The infant was kept in an incubator for 19 days. Muscular hypertonia was observed during the first days of life; this changed to hypotonia at four months. The cardiac pathologies were initially treated with medication (loop diuretics and potassium-sparing diuretics), and the patient was under the care of a cardiologist until reparative surgery was done at 20 months at the Heart Institute in Porto Alegre, RS.

When she was two months old, the patient was referred to the Genetic Counseling Service of the Genetics Department of the Federal University of Pelotas to investigate her neuropsychomotor retardation, which was considered severe. She was evaluated and the diagnosis of trisomy 13, with karyotype 47, XX + 13, was confirmed by chromosomal study. Cytogenetic analyses were done only on cultured peripheral blood lymphocytes. The mother informed that the girl had a convulsive crisis, followed by a cardiac arrest, with spontaneous remission and without receiving medical care, at the fifth month of age. The patient joined the Handi-

capped Parents and Friends Association (APAE) in the city of Canguçu, RS, when she was seven months old. Since then, she has been attending motor physiotherapy sessions weekly. The physiotherapist informed that psychomotor development and muscular hypotonia evolution have been satisfactory. The child is able to maintain herself in a sitting position, roll over and firm the head and shoulders when prone, with little help. She also is able to hold objects, exchanging them from one hand to the other and taking them to her mouth.

The mother noticed that after the beginning of physiotherapy the child was calmer. Her sleep pattern improved and her crying, which had been nearly constant, diminished. It was also noticed that the patient became more receptive and active. She smiles to other people, babbles a few words, and fixes her sight towards objects or sounds that interest her.

On physical examination at 18 months old, the child weighed 6,900 g, had a cephalic perimeter of 41 cm, a thoracic perimeter of 43 cm and was 76 cm tall. At 28 months, she weighed 10,760 g and was 88.5 cm tall (Figures 1 and 2). The face had a low nasal bridge and prominent glabellae. She had ocular hypertelorism, pseudostrabismus and sunken eyes. She also had low-set ears and alopecia in four circular foci at the meeting point of the lambdoid and sagittal sutures. The child had four upper and three lower incisive teeth, but had difficulty in chewing solid food. She ate only liquid and semi-solid food. Pulmonary auscultation revealed a vesicular murmur and no pathological breathing sounds. The precordial examination revealed a regular cardiac rhythm with normophonetic sounds, cardiomegaly and a continuous holopericardiac murmur (+++/++++++), without irradiation. The abdomen was globous, with a moderate umbilical herniation, bowel sounds and absence of hepatomegaly and splenomegaly. The external genitalia were typically feminine, without any detectable abnormality at inspection. The hands had deep palmar creases. She had rocker-bottom feet, with convex soles, and syndactyly of the third and fourth fingers in both feet, with retraction of the second finger. The child could hardly sit without support and did not crawl, but she could raise her head and shoulders when lying face down, looking attentively in the direction of a sound source and could stammer a few syllables. In the skin examination, there was a verrucous lesion on the antero-lateral region of the external malleolus of the right leg (9 mm in diameter) and numerous hemangiomas, including on the internal face of the medial portion of the right forearm (15 mm), the anterior face of the superior third of the left leg (10 mm), left superior-lateral quadrant of the gluteus (5 mm), the left infrascapular region (8 mm), the anterior face of the left shoulder, and the dorsal face of the distal phalange of the third finger of the left hand (2 mm).

Figure 1. The Patau syndrome patient at 18 months.

Figure 2. The Patau syndrome patient at 28 months.

DISCUSSION

The Patau syndrome phenotype typically includes severe central nervous system malformations, such as holoprosencephaly and arinencephaly, with consequent severe psychomotor dysfunction and convulsions (Thompson and Thompson, 1993). This trisomy is characterized by the following triad: microphthalmia, cleft lip and palate and polydactyly. The face may also be characterized by prominent glabellae, ocular hypertelorism, anophthalmia, and micrognathia (Borges-Osório and Robinson, 2001). Our patient had prominent glabellae, ocular hypertelorism and low-set ears. Cleft lip and palate, which are present in 80% of this syndrome's carriers, were absent. The child also did not have micrognathia, micro- or anophthalmia. The observed cardiac malformations, cardiomegaly and perimembranous inter-ventricular communication are consistent with literature descriptions, which indicate that 80% of the cardiac malformations are defects in the inter-ventricular septum. Hemangiomas in diverse regions of the body have also been reported (Schinzel and Gruyler, 1984; Jones, 1998).

Some of the osteomuscular abnormalities reported in the literature (Baraitser and Winter, 1996) were present in our patient, including post-axial polydactyly in the hands, deep palmar creases in the hands, rocker-bottom feet, and convex soles.

Some neurological characteristics of this syndrome, such as spasticity, which changed with time to hypotonia (Carakushansky, 2001), were registered. The alopecia in four circular foci in the lambda region was due to the scars of the imperfect closure of the neural tube, a common finding in trisomy 13 carriers. Neuropsychomotor development retardation was noticeable since birth and, according to the psychologist and the social assistant of APAE-Canguçu, an improvement was observed after the beginning of physiotherapy and recreational sessions at their institution.

The psychological support offered to the mother was, and has been, of great help to the development and maintenance of the mother-child relationship. This close relationship, added to

the tireless maternal effort to provide the best quality of life to her daughter, seems to have contributed positively to the long survival of this child, bypassing the first two years of life.

ACKNOWLEDGMENTS

The authors are thankful to the directors and to the technical staff of APAE in Canguçu, for their help during this study.

REFERENCES

Baraitser, M. and **Winter, R.M.** (1996). *Color Atlas of Congenital Malformation Syndromes*. Mosby-Wolfe, London, England.

Best, R.G. (2002). Patau syndrome. On-line article: http://www.emedicine.com/PED/topic1745.htm. Accessed January 14, 2003.

Borges-Osório, M.R. and **Robinson, W.M.** (2001). *Genética Humana*. Artes Médicas, Porto Alegre, RS, Brazil.

Carakushansky, G. (2001). *Doenças Genéticas em Pediatria*. Guanabara Koogan S.A., Rio de Janeiro, RJ, Brazil.

Goel, M. and **Rathore, R.** (2000). Trisomy 13 (Patau syndrome). *Indian Pediatr. 37*: 1140.

Jones, K.L. (1998). *Padrões Reconhecíveis de Malformações Congênitas*. Editora Manole Ltda., São Paulo, SP, Brazil.

Patau, K., Smith, D.W., Therman, E., Inhorn, S.L. and **Wagner, H.P.** (1960). Multiple congenital anomaly caused by an extra autosome. *Lancet 1*: 790-793.

Schinzel, A.S. and **Gruyler, W.** (1984). *Catalogue of Unbalanced Chromosome Aberrations in Man*. Berlin, New York, NY, USA.

Singh, K.S.T. (1990). Trisomy 13 (Patau's syndrome): a rare case of survival into adulthood. *J. Ment. Defic. Res. 34*: 91-93.

SOFT Volunteers (2002). Health supervision and anticipatory guidance for infants and children with trisomy 18 and 13: suggested guidelines. The Support Organization for Trisomy 18, 13 and other related disorders. On-line article: http://www.trisomy.org/info/html/health_supervision.htm. Accessed August 5, 2003.

Thompson, J.S. and **Thompson, M.W.** (1993). *Genética Médica*. Guanabara Koogan S.A., Rio de Janeiro, RJ, Brazil.

Tunca, Y., Kandale, J.S. and **Pivnick, E.K.** (2001). Long-term survival in Patau syndrome. *Clin. Dysmorphol. 10*: 149-150.

Wideremann, H.R., Grosse, F.R. and **Dibben, H.** (1980). *Característica das Síndromes em Pediatria*. Editora Manole Ltda., São Paulo, SP, Brazil.