Tzu Chi Medical Journal 26 (2014) 192–194

Contents lists available at ScienceDirect

Tzu Chi Medical Journal

journal homepage: www.tzuchimedjnl.com

Clinical Practice and Therapeutics

Clinical management of a patient with der(1) ins(1;2)(p34.1;q23q35)^{mat}

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ARTICLE INFO

Article history:
Received 30 October 2013
Received in revised form
15 November 2013
Accepted 10 February 2014

A 7-month-old boy presented with multiple congenital abnormalities. He had many dysmorphic features, such as a triangular flat face, inverted bilateral epicanthal folds, curly, prominent frontal hair, flat nasal bridge, upturned nose, webbed neck, small chest wall, syndactyly of the right third and fourth fingers, webbed left third and fourth digits, bilateral simian creases, very white skin color, and undescended testes bilaterally (Fig. 1). Other organ anomalies were whole-brain parenchymal tissue atrophy, corpus callosum dysgenesis, sacral dimpling, and cardiomegaly with atrial septal and ventricular septal defects. Agenesis of the left kidney was also found after whole-body survey. der(1) ins(1;2)(p34.1;q23q35)^{mat} was diagnosed from peripheral blood karyotyping (Fig. 2). A study of the mother's chromosomes revealed a balanced translocation of Chromosomes 1 and 2 [ins(1;2)(p34.1;q23q35); Fig. 3].

The patient received medication for epilepsy and mild congestive heart failure. Oral gastric tubing was indicated because of poor sucking and swallowing abilities. An early infant stimulation program was arranged because of severe delay in gross and fine motor milestones, sound making, poor eye contact, and interaction with people and surrounding environment. Developmental milestones and vital organ function should be closely and continuously monitored. Surgical correction of syndactyly and cryptorchidism, regular vaccination, a health supervision program, and an interprofessional patient care program were initiated.

We arranged psychological support for the parents and all family members, including social welfare allocation and genetic counseling. The whole genetic counseling process included explanation of chromosomes and the concept of aneuploidy with the particular chromosomes involved in this case. We educated them about how chromosomal anomalies occur, the mode of inheritance, and the risks in further pregnancies. The outcome could be predicted as follows: 25% chance of the del2(q23q35) genotype, which may result in intrauterine fetal death or spontaneous abortion; 25% chance of the same chromosome anomaly as the proband, der(1) ins(1;2)(p34.1;q23q35)^{mat}, resulting in multiple congenital anomalies; 25% chance the fetus would have the same chromosome anomaly as the mother with no definite major systemic illnesses; and 25% chance of having a normal baby without a chromosome anomaly (Fig. 4).

A fact sheet was not available for the parents because no similar cases were found in a literature review. Instead, available testing options were offered to the parents to prevent another event. These included preimplantation genetic diagnosis, or early prenatal diagnosis such as karyotyping of cord blood and amniocytes in future pregnancies. We emphasized the importance of neonatal screening if there was no prenatal study. The whole process was supportive, nondirective, and nonjudgmental. We emphasized that the chromosomal anomaly had nothing to do with the parents, to ease pressure on them, and all decisions were made based on the parents' personal, cultural, ethical, and social beliefs.

Conflicts of interest: none.

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http://dx.doi.org/10.1016/j.tcmj.2014.03.001

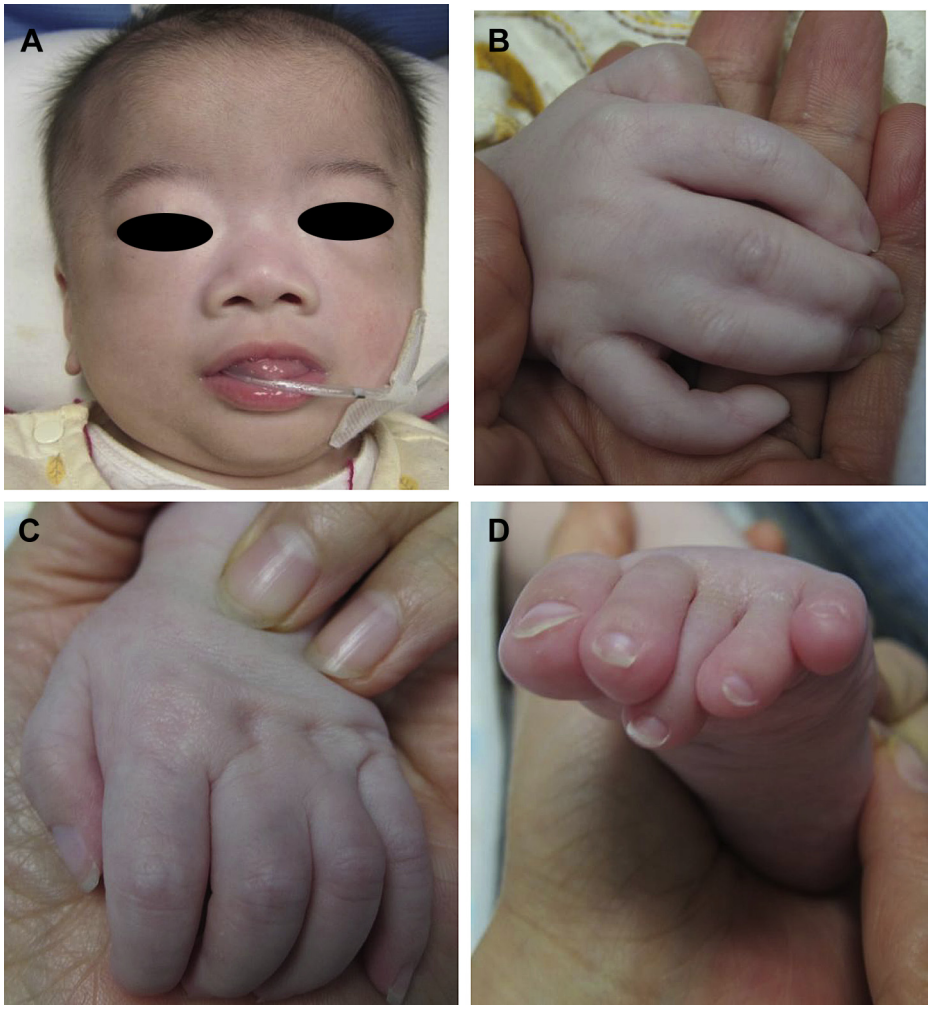

Fig. 1. (A) Dysmorphic features include a triangular, flat face, inverted bilateral epicanthal folds, flat nasal bridge and upturned nose, protruding tongue, and hypopigmented skin. (B) Right third and fourth finger syndactyly. (C) Webbed left third and fourth fingers. (D) Overlapping toes (with parents' permission for publication).

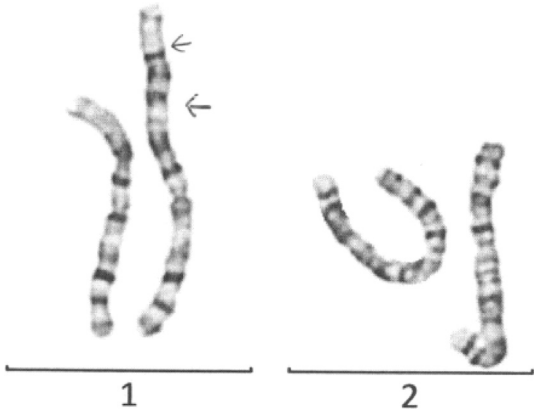

Fig. 2. Chromosomes 1 and 2 of the patient. Arrows indicate der(1) ins(1;2)(p34.1;q23q35).

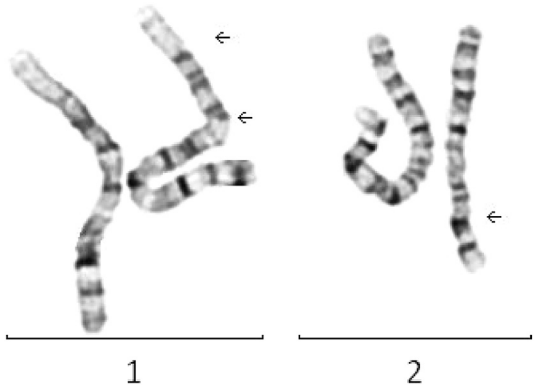

Fig. 3. Chromosomes 1 and 2 of the patient's mother. Arrows indicate ins(1;2)(p34.1;q23q35).

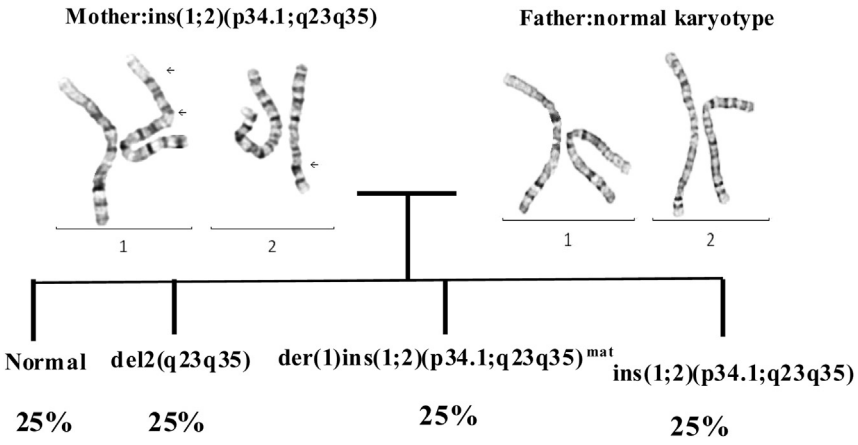

Fig. 4. Chance of 25% of each of the four genotypes in future offspring. Normal, del2(q23q35), der(1)ins(1;2)(p34.1;q23q35)^{mat}, and ins(1;2)(p34.1;q23q35).

Further reading

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