Case Report

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# Partial deletion of the long arm of chromosome 7: a case report

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**Abstract**: Study advances with a childhood case of partial deletion of the long arm of chromosome 7. The patient is a 36-month-old girl with growth retardation, mild mental retardation and delayed bone age. She showed no signs of hypotelorism, upslanting palpebral fissures, epicanthal folds, low-set ears, or flat and broad nasal bridge. Microarray testing using the Affymetrix CytoScan HD array revealed an approximately 58 kb deletion at 7q31.1 in the girl and her father, suggesting paternal origin. As the patient had no characteristic facial features, 7q deletions had not been considered. This case broadens the range of case presentations for microdeletions of chromosome 7.

**Keywords:** Chromosome 7; Partial deletion of the long arm; Growth retardation

## 1 Introduction

Chromosome microdeletion is a hereditary disease with complex clinical manifestations due to the deletion of small chromosome segments [1]. The length of chromosome deletion is less than 5Mb. The common clinical manifestations include: growth and development abnormalities, mental retardation, organ malformations, unusual facies, endocrine abnormalities, mental and behavioral changes, etc. [2]. Chromosome 7 contains more than 158 million base pairs and represents approximately 5% of total cellular DNA [3]. Partial deletion of the long arm of chromosome 7 is a rare chromosomal variant. The first cases of partial deletion of the long arm of chromosome 7 were reported in 1968 [4], and many cases have been reported since. The deletion of chromosome 7 is more common in q32→qter, and deletions in the q31 region are rarely reported. In this publication, we report a case of chromosome 7 with a 58 kb deletion in the paternal q31.1 segment.

## 2 Case presentation

A 36-month-old girl presented to our service. She had been diagnosed with intrauterine growth restriction (birth weight 1.98 kg, height 44 cm) at 38 weeks' gestation. Echocardiography after birth revealed a patent ductus arteriosus and an atrial septal defect. Her parents denied familial disease or history of infection. No substantial dysmorphic features were observed. At 4-months of age, she presented to our hospital for high-risk infant follow-up. In the process of follow-up physical examination, we found that her weight, height, and head circumference were all below the 3rd percentile. A urinary galactose test was positive. Her aspartate aminotransferase (AST) was 274.4 U/L, and her alanine aminotransferase (ALT) was 266.6U/L, both above the range of normal. We referred the family to the Gastroenterology department for further management. When the patient was followed up at 5-months of age, her AST and ALT were normal. At 6-months of age, it was observed that the girl did not exhibit stranger anxiety. She had difficulty with hand-eye coordination. She was unable to reach for objects with one hand or to grab objects. Gesell examination suggested developmental delay. We provided her parents with guidance regarding cognitive training to use at home. The infant began babbling and learning by imitation at 10 months of age. During a 20-item neuromotor assessment, we found that dorsiflexion angles were asymmetric in both feet. We provided motor functional training and instruction to sup-

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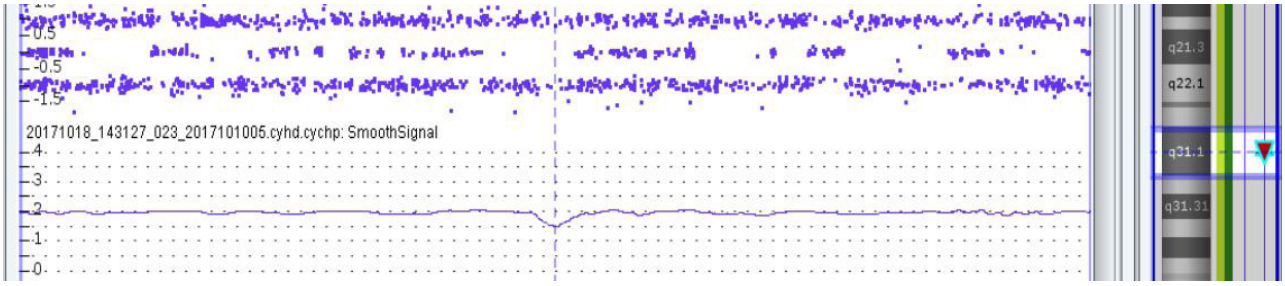


**Figure 1:** The patient's aCGH analysis chart, in which the arrowhead refers to the part of the patient's 58 kb deletion at 7q31.1.

plement cognitive training. Her patent ductus arteriosus was surgically corrected at 12 months of age. At 18 months of age, her language was equivalent to that of a 12-month-old. Her receptive and expressive speech improved by 22 months of age, after speech-language training.

Chromosomal evaluation of the patient and her parents using the Affymetrix CytoScan HD array revealed an approximately 58kb deletion at 7q31.1 in the girl (Fig.1) and her father, suggesting that the deletion was of paternal origin. An electroencephalogram (EEG) was normal. Brain MRI was also normal. No abnormalities were detected by tandem mass spectrometry and bone age was delayed (> 2 SD).

**Ethical approval:** The research related to human use has been complied with all the relevant national regulations, institutional policies and in accordance the tenets of the Helsinki Declaration, and has been approved by the authors' institutional review board or equivalent committee.

**Informed consent:** Informed consent has been obtained from patient included in this study.

## 3 Discussion

Harris *et al* [5] in their research, summarized the common clinical manifestations of partial deletion of long arm of chromosome 7, including retardation of growth and motor development, mental retardation, hypotelorism, upslanting palpebral fissures, epicanthal folds, low-set ears, flat and broad nasal bridge, abnormal muscular tension and, feeding problems. This patient's clinical history was remarkable for pre- and postnatal growth retardation, mental retardation and delayed bone age. These findings are consistent with a report of partial deletion of the long arm of chromosome 7 [6]. The diagnosis of 7q deletion syndrome was definitively established by gene chip analysis.

Growth retardation was the most significant feature of this case. In retrospect, the following factors were salient: 1) intrauterine growth retardation. Children with intrauterine growth retardation usually achieve catch-up growth prior to age 2, though in some reports approximately 10%-20% of intrauterine growth retarded children have persistent growth delay after birth [7,8]; 2) growth delay despite correction of lactose intolerance: unlike adults, children with severe lactose intolerance may develop chronic diarrhea, weight loss, and a severe kwashiorkor-like syndrome. Although the patient's urine galactose test was positive, there were no typical symptoms of lactose intolerance: bloating, diarrhea, and abdominal pain [9]. In addition, diagnosing lactose intolerance is difficult because it depends on self-reported symptoms that are variable and thus may not be assessed objectively [10]. The patient was given intermittent lactase preparations, with no significant improvement in weight gain; 3) congenital heart disease: patent ductus arteriosus and atrial septal defect. The child's patent ductus arteriosus was repaired at 12 months of age. She has been scheduled for evaluation for repair of her atrial septal defect.

The genetic research of chromosome microdeletion mainly focuses on the localization of pathogenic genes and functional analysis of gene loci. Previous studies have shown that IMMP2L, DOCK4, GPR85 and Foxp2 gene are located at 7q31.1, which are closely related to nervous system development [11]. For example, Maestrini et al [12] reported that IMMP2L and DOCK4 gene are likely to be associated with autism. Moreover, Hellebrand et al [13] found that in the central nervous system Gpr85 was expressed during phases of early neuronal differentiation and play a specific function for differentiation processes in the cerebral cortex. FOXP2 gene is the first language related gene discovered by human beings. In the development of neural circuits related to language learning, FOXP2 and its downstream target genes may constitute a decisive gene network [14]. Despite decades of research, the relationship between genotype and phenotype of the partial deletion of the long arm of chromosome 7 is

still unclear [15]. The finding of a chromosomal deletion is only the beginning of this investigation. It is necessary to search for missing features, to locate out the precise breakpoints, and to study the genes involved and evaluate the causes of the phenotypic variation. This will be the focal point of a future next study.

It is important to appreciate the association of clinical phenotypes and genes in microdeletions of this chromosomal region. Once the syndrome is recognized, active interventions should be carried out for long periods to improve the quality of life of affected children.

## 4 Conclusion

In conclusion, this case indicated an impressive clinical diagnosed as partial deletion of the long arm of chromosome 7 syndrome by gene analysis. As the patient had no characteristic facial features, the report can provide more information for clinic diagnosis and for growth retardation.

**Competing interests:** The authors declare that they have no competing interests.

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**Authors' Contribution:** Chun Zhu collected data, drafted the manuscript and revised the paper.Mei-Ling Tong monitore data collection for the whole process and revised the paper. Xia Chi designed the study. She is responsible for the integrity of this study.

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