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Case Report

A New Case of Doors Syndrome in China

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Abstract

DOORs syndrome is a rare disorder, with its acronym derived from a syndromic presentation of Deafness, Onychodystrophy, Osteodystrophy, mental Retardation and seizures. Its aetiology is yet fully understood. DOORs syndrome has previously been reported worldwide but not in the ethnic Chinese population. As one of the biggest general hospitals in South West China, we have encountered two cases resembling DOORs syndrome in recent practice, it is the first time to report the disorder in China.

Introduction

DOORs syndrome is a rarely described disorder which includes sensorineural hearing loss, hypoplastic or absent nails on the hands and feet, small or absent distal phalanges of the hands and feet, and mental retardation. DOORs is an acronym for the 4 main symptoms. We report two cases whose clinical manifestations accord with DOORs syndrome. which are presented and discussed below.

Conclusion

Case 1

The first patient was a 4-year-old boy who presented to the Ear, Nose and Throat department with bilateral sensorineural hearing loss. We learnt from his parents that the patient was conceived and delivered naturally by his young parents, both in their 20s, through a non-consanguineous marriage. He was the first child and there was no family history of any intellectual disability, seizures, congenital defects or genetic disorders. Physical examination revealed retarded growth with his weight (14kg, <16kg), height (96cm, <103cm) and head circumference skull circumference(48cm,) well below the national average. Musculoskeletal examinations revealed His fifth finger were shortened, no nails on bilaterally thumbs and the nails on fifth fingers were markly rudimentary while others were slightly rudimentary. All of the nails on his toes were absent(Fig.1). In addition, he exhibited severe psychomotor retardation and has not yet acquired any language skills aged 4. Pure tone audiogram showed bilateral sensorineural hearing loss (Fig 2A). Tympanogram showed type A pattern in both ears. Otoacoustic Emission test (OAE) was negative. Click-evoked Auditory Brainsteam Response (ABR) was only obtained at a threshold of 75dBnHL by monoaural stimulation through a headphone.

Case 2

The second patient was a 6-year-old boy, who was admitted to our department because of hearing loss. He began to speak when he was 2 years old, but indistinct. In the past 2 years, his parents found the boy responded to sound (the voice of TV or talking) less than before. The parents were not consanguinity, and the boy was born from the first pregnancy and when his parents were over 20 years old.

There was no response to the otoacoustic emission. The threshold of auditory brainstem response was 45dBnHL by the monaural stimulation with alternate clicks through a headphone(Fig.2B).

Unfortunately, neither of the children received any further pediatric evaluation or genetic testing due to the financial hardship of the families, both of which were from remote rural China far from the hospital's catchment area. This makes any treatment or follow-up planning challenging for the families. In the end, both families declined any further medical input to their children. We felt disheartened that a good opportunity to thoroughly study this rare syndrome in the Chinese population was lost.

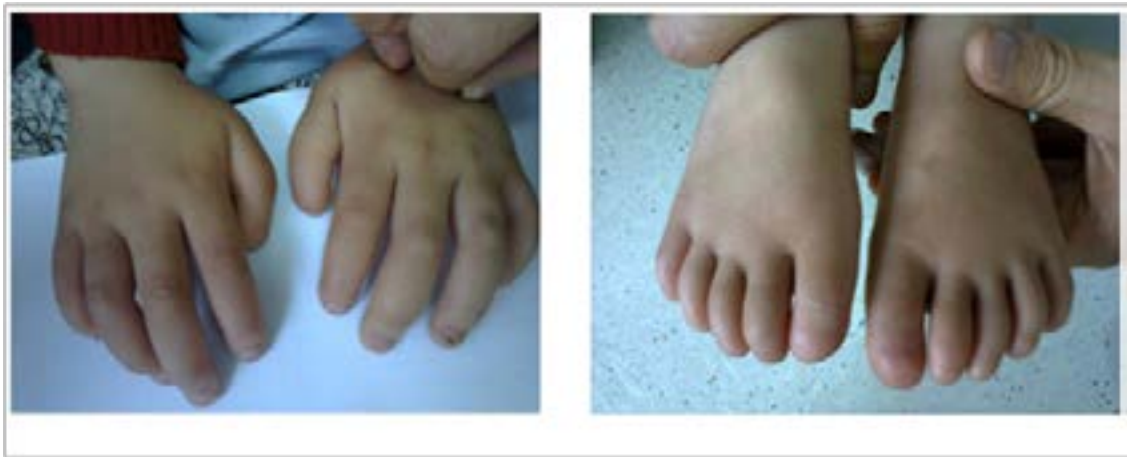

Figure.1: No nails on bilaterally thumbs and the nails on fifth fingers were markly rudimentary while others were slightly rudi mentary. All of the nails on his toes were absent.

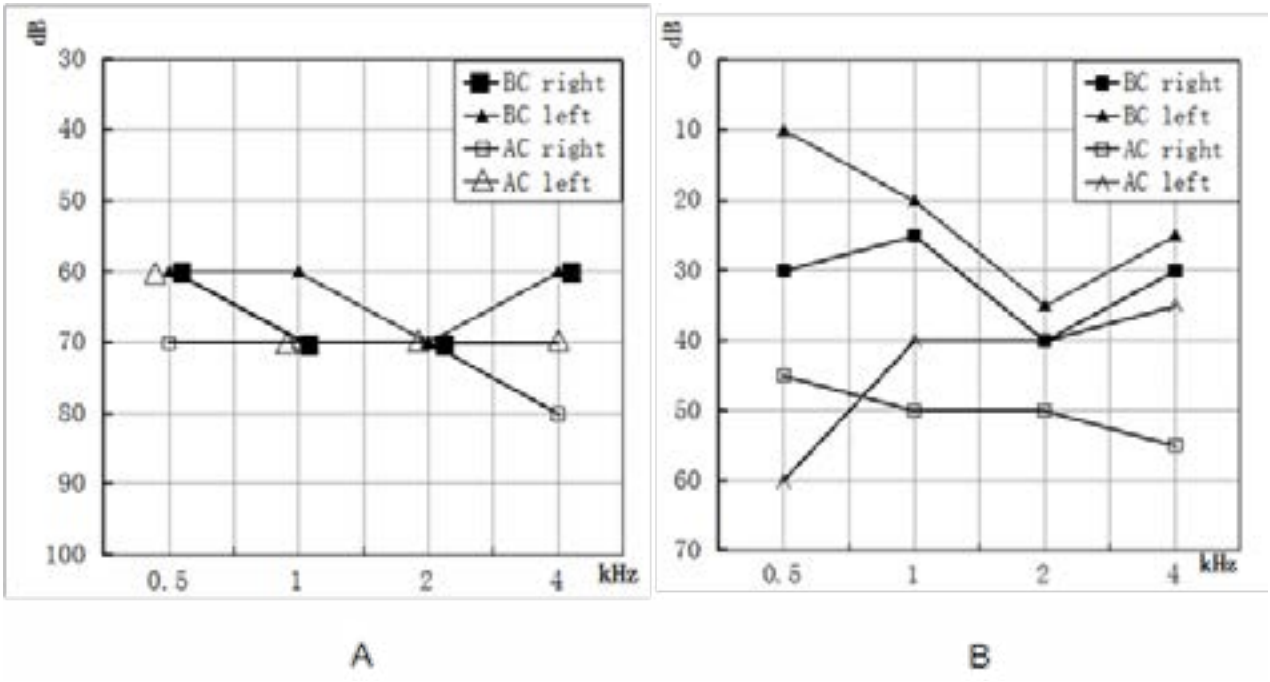

Figure.2: Air conduction (AC) and bone conduction (BC) thresholds for the two children.

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