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Microdeletion on 17p11.2 in a Smith-Magenis syndrome patient with mental retardation and congenital heart defect: first report from China

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ABSTRACT. Smith-Magenis syndrome (SMS) is a rare syndrome with multiple congenital malformations, including development and mental retardation, behavioral problems and a distinct facial appearance. SMS is caused by haploinsufficiency of *RAI1* (deletion or mutation of *RAI1*). We describe an eight-year-old female Chinese patient with multiple malformations, congenital heart defect, mental retardation, and behavioral problems (self hugging, sleeping disturbance). High-resolution genome wide single nucleotide polymorphism array revealed a 3.7-Mb deletion in chromosome region 17p11.2. This chromosome region contains *RAI1*, a critical gene involved in SMS. To

the best of our knowledge, this is the first report of an SMS patient in mainland China.

Key words: Smith-Magenis syndrome; Congenital heart defect; High-resolution single nucleotide polymorphism; Sleeping disturbance; Mental retardation

