A compound heterozygote mutation in a Chinese patient affected with Methylmalonic acidemia

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Objectives:

The aim of this study was to detect potential gene mutation of Methylmalonic acidemia (MAA) in a Chinese patient.

Methods:

Patient with clinical diagnosis and parents were analyzed in this study. The analysis included medical histories, clinical analysis, and genetic tests. A NGS panel include MUT,MMAA,MMAB,MMADHC and MCEE genes was identify the pathogenic mutation responsible for the MAA and verified by Sanger.

Results:

A compound heterozygote mutation c.571C>T（p.R191W）and c.539C>G（p.S180W）of the MMAB gene was found in the patient, and inherit from his father and mother. The same mutations were not found among 100 healthy controls.

Conclusions:

A compound heterozygote mutation c.571C>T（p.R191W）and c.539C>G（p.S180W）of the MMAB gene mutation can be a cause of Cb1b MAA in Chinese. We think that genetic studies to may assist in making Cb1b MAA diagnosis and providing the consultant for their families.