A novel homozygous mutation in ERCC8 cause Cockayne Syndrome a in a Chinese family

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Objectives:

Short stature can be caused by mutations in a multitude of different genes. Cockayne Syndrome is a rare growth disorder marked by progressive growth failure, neurologic abnormality. The current report describes a patient with severe short stature and neurologic abnormality.

Methods:

Patient with clinical diagnosis and parents were analyzed in this study. The analysis included medical histories, clinical analysis, and genetic tests. The gene was sequenced to identify the pathogenic mutation responsible for the development of Cockayne Syndrome by qPCR.

Results:

Inherited disease panel identified a novel homozygous mutation c.394_398delTTACA in ERCC8 that had not been previously reported. qPCR analysis revealed c.394_398delTTACA was maternal and his father was heterozygous.

Conclusions:

A novel homozygous mutation c.394_398delTTACA in ERCC8 gene can be a cause of Cockayne Syndrome in Chinese. The novel mutations have enriched the mutation spectrum of the ERCC8 gene.