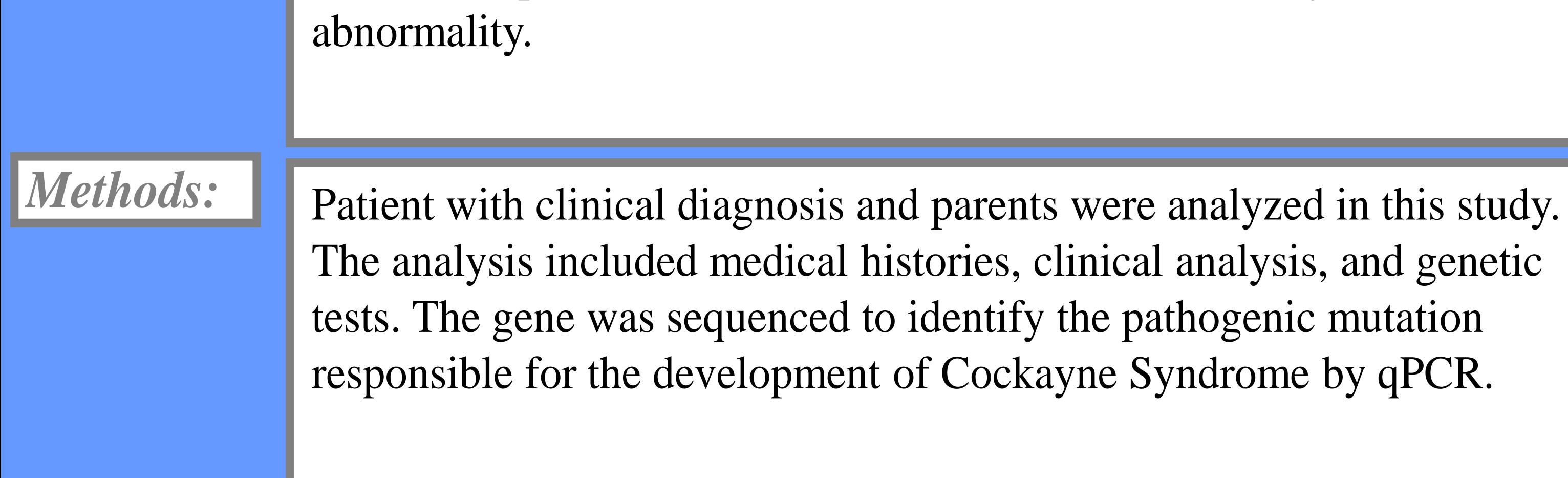
A novel homozygous mutation in ERCC8 cause Cockayne Syndrome a in a Chinese family

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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





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.



Hui Huang

Growth and syndromes (to include Turner syndrome)





