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## Large-single scale mitochondrial DNA deletions in different tissues of patients with Kearns-Sayre syndrome

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**Objective:** To investigate the clinical significance of samples except skeletal muscle that could be detected the large-scale single deletions directly in the diagnosis of Kearns-Sayre syndrome by concluding the clinical and genetic features of KSS.

**Methods:** The clinical data of four patients with KSS, which were diagnosed from 2016 to 2017 by Beijing Children's Hospital, Capital Medical University, were collected. The clinical features and gene mutation characteristics were analyzed retrospectively.

**Results:** 4 patients were all consistent with the diagnosis criteria of KSS, the age of onset was 8.2 years old on average (5.0-11.8), and the initial symptoms were no specificity. The common symptoms of the four cases were exercise intolerance, short statue, ophthalmoplegia, hypotonia, muscle weakness; cerebrospinal fluid protein concentration over 1000mg/L, the cerebral MRI showed that abnormal signals in the brainstem, in addition, white matter, thalamus, basal ganglia, cerebrum and cerebellum atrophy could be found. Moreover, three cases for cardiac conduction block, two for suspicious family history. Molecular analysis of the four cases revealed that the large-scale single deletions of mitochondrial DNA from the peripheral blood, the urine and other samples (muscle tissue, CSF, etc.) through the next-generation sequencing. Two cases for homogeneous variation. Of three cases who did pedigree analysis, only the mom of case 4 was detected the same variation with the proband.

**Conclusions:** With the development of the next-generation sequencing, the diagnosis of Kearns-Sayre syndrome was no longer than depending on the muscle biopsy. It is indeed possible to detect the large-scale single deletions in peripheral blood, urine and other samples, to improve the molecular diagnosis of KSS, which will have a significant influence on the diagnosis and management of KSS, but more cases will be needed in the future.

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### Biography

Yuqing shi is a master student major in children neurology of professor FangFang, Beijing Children's Hospital, who has her expertise in evaluation and passion in improving the life quality of the patients and their families with mitochondrial diseases.

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