GENETICS AND GENODERMATOSES

ROTHMUND-THOMSON SYNDROME: 4 NEW CASES FROM THREE UNRELATED FAMILIES IN CHINA

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Background: Rothmund-Thomson syndrome (RTS) is a rare autosomal recessive disorder characterized by facial poikiloderma, growth retardation, sparse scalp hair/eyelashes/eyebrows, juvenile cataracts, skeletal abnormalities, radial ray defects and a predisposition to cancer. Around 400 cases have been reported in the literature to date. The mutations in the RecQ-like DNA helicase type 4 (RECQL4) gene can be found in approximately 65%–70% of RTS cases and diagnosis needs to based on a combination of clinical and gene sequencing.

Observation: We herein reported 4 new cases from three unrelated chinese families, two of them are siblings, and highlighted the variability of the clinical presentation with or with not RECQL4 gene mutation. Clinical data of 4 Chinese children with classical features of RTS were collected. Proband and family members' blood samples were obtained to analyze their RECQL4 gene variants using Illumina NextSeq platform. All probands presented with poikiloderma on the face within their first 2 years of age. In addition, growth delay, sparse or absent eyebrows, teeth retardation and toe deformity were noted in two probands. One of them presented epicanthus. Juvenile cataract described in previously reported RTS cases hadn't been detected in our probands. Two novel RECQL4 variants were identified in one of the probands, including a point mutation (c.1391-2A>C) producing a splice acceptor variant and a deletion of two nucleotides (p.His831Argfs) producing a frame shift.

Key message:
Rothmund-Thomson syndrome (RTS) is a rare autosomal recessive disorder characterized by facial poikiloderma, growth retardation and RECQL4 gene mutation. Our study further expands clinical features of RTS and reveal novel phenotypes observed in Chinese RTS patients.