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Multisystemic smooth muscle dysfunction syndrome

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Multisystem smooth muscle dysfunction syndrome in China: A case report

Sai-Nan Chen, Yu-Qing Wang, Chuang-Li Hao, Yan-Hong Lu, Wu-Jun Jiang, Chun-Yan Gao, Min Wu

Abstract

BACKGROUND

Multisystemic smooth muscle dysfunction syndrome (MSMDS) is a rare genetic disease worldwide. The main mutation is the actin alpha2 (ACTA2) gene p.R179H. In this study, we aimed to report a Chinese MSMDS patient and systematically review the previous literature.

CASE SUMMARY

Here, we report a 9.6-month-old Chinese girl who was diagnosed with MSMDS based on her history and symptoms, such as recurrent cough, wheezing and complications with congenital fixed dilated pupils. Chest high-resolution computed tomography scanning

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Smooth muscle cells (SMCs) contract to perform many physiological functions, including regulation of **blood flow** and pressure in arteries, contraction of the pupils, peristalsis of the gut and voiding of the bladder. SMC lineage in these organs is characterized by cellular expression of the SMC isoform of **α-actin**, encoded by the ACTA2 gene.

Cited by: 134 **Author:** Dianna M. Milewicz, John R. Østergaard, ...

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Cited by: 2 **Author:** Hui Li, Yumeng Huang, Yan Li, Baozhong...

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