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| Dear Reader,We are pleased to deliver your requested table of contents alert for **Science China Life Sciences**. [**Volume 60 Number 7**](https://urldefense.proofpoint.com/v2/url?u=http-3A__alerts.springer.com_re-3Fl-3DD0In67cwjI6gyt153Iy&d=DwMFaQ&c=ZQs-KZ8oxEw0p81sqgiaRA&r=rOMURTZ4fslq9dKzVxltN4xNU-D2Ru1wiL30akyNmwk&m=nj0nvs80NSedvgnUVjyqeFA-89KOaGWZHHZ_lYdP2B0&s=QjKnQ_b9Eyt7_lSbdCdhfXmcQC14lUpa1J_yOBq17Zs&e=) is now available online.Thematic Issue: Frontiers in rare diseases |  |

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| In this issue |
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| Editorial |  |

The challenge and promise of rare disease diagnosis in ChinaXin Ni & Tieliu Shi |

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| Review |  |

Towards efficiency in rare disease research: what is distinctive and important?Jinmeng Jia & Tieliu Shi |

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| Research Paper |  |

Genome-wide analysis of differential DNA methylation in Silver-Russell syndromeDi Wu, Chunxiu Gong & Chang Su |

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| Research Paper |  |

*AR* mutations in 28 patients with androgen insensitivity syndrome (Prader grade 0–3)Yi Wang, Chunxiu Gong, Xiou Wang & Miao Qin |

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| Research Paper |  |

Clinical feature and waveform in infantile nystagmus syndrome in children with *FRMD7* gene mutationsDayong Bai, Wei Shi, Zhan Qi, Wei Li, Aihua Wei, Yanhui Cui, Cheng Li & Li Li |

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| Research Paper |  |

*DICER1* mutations in twelve Chinese patients with pleuropulmonary blastomaSiyu Cai, Xisi Wang, Wen Zhao, Libing Fu, Xiaoli Ma & Xiaoxia Peng |

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| Research Paper |  |

Detection of *FOXO1* break-apart status by fluorescence *in situ* hybridization in atypical alveolar rhabdomyosarcomaLibing Fu, Yaqiong Jin, Chao Jia, Jie Zhang, Jun Tai, Hongbin Li, Feng Chen, Jin Shi, Yongli Guo, Xin Ni & Lejian He |

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| Research Paper |  |

Correlation between *BRAF* V600E mutation and clinicopathological features in pediatric papillary thyroid carcinomaJiangqiao Geng, Huanmin Wang, Yuanhu Liu, Jun Tai, Yaqiong Jin, Jie Zhang, Lejian He, Libing Fu, Hong Qin, Yingluan Song, Jinzhu Su, Aiying Zhang, Xin Wen, Yongli Guo & Xin Ni |

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| Research Paper |  |

Whole-exome sequencing identified compound heterozygous variants in*MMKS* in a Chinese pedigree with Bardet-Biedl syndromeZhan Qi, Ying Shen, Qian Fu, Wei Li, Wei Yang, Wenshan Xu, Ping Chu, Yaxin Zhang & Hui Wang |

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| Research Paper |  |

The clinical and genetic characteristics in children with mitochondrial disease in ChinaFang Fang, Zhimei Liu, Hezhi Fang, Jian Wu, Danmin Shen, Suzhen Sun, Changhong Ding, Tongli Han, Yun Wu, Junlan Lv, Lei Yang, Shufang Li, Jianxin Lv & Ying Shen |

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| Research Paper |  |

Gene mutations and clinical phenotypes in Chinese children with Blau syndromeCaifeng Li, Junmei Zhang, Shipeng Li, Tongxin Han, Weiying Kuang, Yifang Zhou, Jianghong Deng & Xiaohua Tan |

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| Research Paper |  |

Analysis of genotypes and phenotypes in Chinese children with tuberous sclerosis complexDayong Bai, Junyang Zhao, Li Li, Jun Gao & Xu Wang |

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| Letter to the Editor |  |

Berry syndrome: a rare cardiac malformation with extra-cardiac findingsJingya Li, Ya Yang, Xiaomin Duan, Lanzhong Jin, Lin Zheng, Xin Zhang, Haiyan Wei, Yan Sun, Xiaolin Zhang, Pei Li, Jiao Yang, Ning Ma & Fangyun Wang |

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| Letter to the Editor |  |

Detection of mycobacterial and viral DNA in Kikuchi-Fujimoto disease: an analysis of 153 Chinese pediatric casesZhe Xu, Ying Liu, Haijing Li, Shufang Meng, Alan S. Boyd, Charles W. Stratton, Lin Ma & Yi-Wei Tang |

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| Letter to the Editor |  |

Analysis of diagnosis and treatment of lipoblastomatosisZhiqiang Mo, Xianghui Xie, Huanmin Wang, Hong Qin, Wei Han & Xiaosong Li |

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