|  |  |  |
| --- | --- | --- |
| |  |  | | --- | --- | | 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 |  | |
| In this issue |
| |  |  |  | | --- | --- | --- | | |  |  | | --- | --- | | Editorial |  |   The challenge and promise of rare disease diagnosis in China  Xin Ni & Tieliu Shi | |
| |  |  | | --- | --- | | |  | | --- | | [**» Abstract**](https://urldefense.proofpoint.com/v2/url?u=http-3A__alerts.springer.com_re-3Fl-3DD0In67cwjI6gyt153I10&d=DwMFaQ&c=ZQs-KZ8oxEw0p81sqgiaRA&r=rOMURTZ4fslq9dKzVxltN4xNU-D2Ru1wiL30akyNmwk&m=nj0nvs80NSedvgnUVjyqeFA-89KOaGWZHHZ_lYdP2B0&s=pvyjhPfL8PpmjPW-acQsrl4S3uYq3Fd1ZpPSBhBRGzg&e=)   [**» Full text PDF**](https://urldefense.proofpoint.com/v2/url?u=http-3A__alerts.springer.com_re-3Fl-3DD0In67cwjI6gyt153I11&d=DwMFaQ&c=ZQs-KZ8oxEw0p81sqgiaRA&r=rOMURTZ4fslq9dKzVxltN4xNU-D2Ru1wiL30akyNmwk&m=nj0nvs80NSedvgnUVjyqeFA-89KOaGWZHHZ_lYdP2B0&s=azGqnb-DGBX9RbSArInBQx9RlQmsB8USVj39O7FfjtE&e=) | | |
| |  |  |  | | --- | --- | --- | | |  |  | | --- | --- | | Review |  |   Towards efficiency in rare disease research: what is distinctive and important?  Jinmeng Jia & Tieliu Shi | |
| |  |  | | --- | --- | | |  | | --- | | [**» Abstract**](https://urldefense.proofpoint.com/v2/url?u=http-3A__alerts.springer.com_re-3Fl-3DD0In67cwjI6gyt153I12&d=DwMFaQ&c=ZQs-KZ8oxEw0p81sqgiaRA&r=rOMURTZ4fslq9dKzVxltN4xNU-D2Ru1wiL30akyNmwk&m=nj0nvs80NSedvgnUVjyqeFA-89KOaGWZHHZ_lYdP2B0&s=6EM8xEZuFH-rKFx-MVRA_YGK2OrCrWqEP0cHrIkqevU&e=)   [**» Full text PDF**](https://urldefense.proofpoint.com/v2/url?u=http-3A__alerts.springer.com_re-3Fl-3DD0In67cwjI6gyt153I13&d=DwMFaQ&c=ZQs-KZ8oxEw0p81sqgiaRA&r=rOMURTZ4fslq9dKzVxltN4xNU-D2Ru1wiL30akyNmwk&m=nj0nvs80NSedvgnUVjyqeFA-89KOaGWZHHZ_lYdP2B0&s=3TD1IX3ZIqzPjj_fbZ8K1MgW6S0-A5u55UBjbiNZAh0&e=) | | |
| |  |  |  | | --- | --- | --- | | |  |  | | --- | --- | | Research Paper |  |   Genome-wide analysis of differential DNA methylation in Silver-Russell syndrome  Di Wu, Chunxiu Gong & Chang Su | |
| |  |  | | --- | --- | | |  | | --- | | [**» Abstract**](https://urldefense.proofpoint.com/v2/url?u=http-3A__alerts.springer.com_re-3Fl-3DD0In67cwjI6gyt153I14&d=DwMFaQ&c=ZQs-KZ8oxEw0p81sqgiaRA&r=rOMURTZ4fslq9dKzVxltN4xNU-D2Ru1wiL30akyNmwk&m=nj0nvs80NSedvgnUVjyqeFA-89KOaGWZHHZ_lYdP2B0&s=MLhcb7KrgiPjeq3VcGH1BUT6NscMWnuzp203IzV85fE&e=)   [**» Full text PDF**](https://urldefense.proofpoint.com/v2/url?u=http-3A__alerts.springer.com_re-3Fl-3DD0In67cwjI6gyt153I15&d=DwMFaQ&c=ZQs-KZ8oxEw0p81sqgiaRA&r=rOMURTZ4fslq9dKzVxltN4xNU-D2Ru1wiL30akyNmwk&m=nj0nvs80NSedvgnUVjyqeFA-89KOaGWZHHZ_lYdP2B0&s=pYcMKZecf0fEpmH-9X9vYeJN5yMS8ZVEHUkWFEbvPYM&e=) | | |
| |  |  |  | | --- | --- | --- | | |  |  | | --- | --- | | Research Paper |  |   *AR* mutations in 28 patients with androgen insensitivity syndrome (Prader grade 0–3)  Yi Wang, Chunxiu Gong, Xiou Wang & Miao Qin | |
| |  |  | | --- | --- | | |  | | --- | | [**» Abstract**](https://urldefense.proofpoint.com/v2/url?u=http-3A__alerts.springer.com_re-3Fl-3DD0In67cwjI6gyt153I16&d=DwMFaQ&c=ZQs-KZ8oxEw0p81sqgiaRA&r=rOMURTZ4fslq9dKzVxltN4xNU-D2Ru1wiL30akyNmwk&m=nj0nvs80NSedvgnUVjyqeFA-89KOaGWZHHZ_lYdP2B0&s=bXe4X3wou7nGvECDzwznjnON3Vn_3MrrpcSHshGS2eU&e=)   [**» Full text PDF**](https://urldefense.proofpoint.com/v2/url?u=http-3A__alerts.springer.com_re-3Fl-3DD0In67cwjI6gyt153I17&d=DwMFaQ&c=ZQs-KZ8oxEw0p81sqgiaRA&r=rOMURTZ4fslq9dKzVxltN4xNU-D2Ru1wiL30akyNmwk&m=nj0nvs80NSedvgnUVjyqeFA-89KOaGWZHHZ_lYdP2B0&s=8gTzagmCMDSAzii4JMUz7-TLU4GRuYfHtgGWF7BEMrg&e=) | | |
| |  |  |  | | --- | --- | --- | | |  |  | | --- | --- | | Research Paper |  |   Clinical feature and waveform in infantile nystagmus syndrome in children with *FRMD7* gene mutations  Dayong Bai, Wei Shi, Zhan Qi, Wei Li, Aihua Wei, Yanhui Cui, Cheng Li & Li Li | |
| |  |  | | --- | --- | | |  | | --- | | [**» Abstract**](https://urldefense.proofpoint.com/v2/url?u=http-3A__alerts.springer.com_re-3Fl-3DD0In67cwjI6gyt153I18&d=DwMFaQ&c=ZQs-KZ8oxEw0p81sqgiaRA&r=rOMURTZ4fslq9dKzVxltN4xNU-D2Ru1wiL30akyNmwk&m=nj0nvs80NSedvgnUVjyqeFA-89KOaGWZHHZ_lYdP2B0&s=1RdesARic4w7iLSuDasFk5CXGAqvlIUZpYeunQuLjtQ&e=)   [**» Full text PDF**](https://urldefense.proofpoint.com/v2/url?u=http-3A__alerts.springer.com_re-3Fl-3DD0In67cwjI6gyt153I19&d=DwMFaQ&c=ZQs-KZ8oxEw0p81sqgiaRA&r=rOMURTZ4fslq9dKzVxltN4xNU-D2Ru1wiL30akyNmwk&m=nj0nvs80NSedvgnUVjyqeFA-89KOaGWZHHZ_lYdP2B0&s=WQkio7TMTHg_2xbQj97IeVcuPGCrJn4dfocmeP-DC1w&e=) | | |
| |  |  |  | | --- | --- | --- | | |  |  | | --- | --- | | Research Paper |  |   *DICER1* mutations in twelve Chinese patients with pleuropulmonary blastoma  Siyu Cai, Xisi Wang, Wen Zhao, Libing Fu, Xiaoli Ma & Xiaoxia Peng | |
| |  |  | | --- | --- | | |  | | --- | | [**» Abstract**](https://urldefense.proofpoint.com/v2/url?u=http-3A__alerts.springer.com_re-3Fl-3DD0In67cwjI6gyt153I1a&d=DwMFaQ&c=ZQs-KZ8oxEw0p81sqgiaRA&r=rOMURTZ4fslq9dKzVxltN4xNU-D2Ru1wiL30akyNmwk&m=nj0nvs80NSedvgnUVjyqeFA-89KOaGWZHHZ_lYdP2B0&s=0AWAX_NkyiOgiYgKCygpep6FSlS4HlOBrIS5n2Avj2o&e=)   [**» Full text PDF**](https://urldefense.proofpoint.com/v2/url?u=http-3A__alerts.springer.com_re-3Fl-3DD0In67cwjI6gyt153I1b&d=DwMFaQ&c=ZQs-KZ8oxEw0p81sqgiaRA&r=rOMURTZ4fslq9dKzVxltN4xNU-D2Ru1wiL30akyNmwk&m=nj0nvs80NSedvgnUVjyqeFA-89KOaGWZHHZ_lYdP2B0&s=r-NRDJoGLl1iIdMK-U-9HWyUJqI3LPQxxNZi_1O7WhQ&e=) | | |
| |  |  |  | | --- | --- | --- | | |  |  | | --- | --- | | Research Paper |  |   Detection of *FOXO1* break-apart status by fluorescence *in situ* hybridization in atypical alveolar rhabdomyosarcoma  Libing Fu, Yaqiong Jin, Chao Jia, Jie Zhang, Jun Tai, Hongbin Li, Feng Chen, Jin Shi, Yongli Guo, Xin Ni & Lejian He | |
| |  |  | | --- | --- | | |  | | --- | | [**» Abstract**](https://urldefense.proofpoint.com/v2/url?u=http-3A__alerts.springer.com_re-3Fl-3DD0In67cwjI6gyt153I1c&d=DwMFaQ&c=ZQs-KZ8oxEw0p81sqgiaRA&r=rOMURTZ4fslq9dKzVxltN4xNU-D2Ru1wiL30akyNmwk&m=nj0nvs80NSedvgnUVjyqeFA-89KOaGWZHHZ_lYdP2B0&s=mBZGi8WES3j_dKVaGLfLu5VXq0u5iYQD7BuXu5myHAA&e=)   [**» Full text PDF**](https://urldefense.proofpoint.com/v2/url?u=http-3A__alerts.springer.com_re-3Fl-3DD0In67cwjI6gyt153I1d&d=DwMFaQ&c=ZQs-KZ8oxEw0p81sqgiaRA&r=rOMURTZ4fslq9dKzVxltN4xNU-D2Ru1wiL30akyNmwk&m=nj0nvs80NSedvgnUVjyqeFA-89KOaGWZHHZ_lYdP2B0&s=8dfh_XkHjGPmnITe8_y0gO6M0seuGTb3uSBrQg1-qWU&e=) | | |
| |  |  |  | | --- | --- | --- | | |  |  | | --- | --- | | Research Paper |  |   Correlation between *BRAF* V600E mutation and clinicopathological features in pediatric papillary thyroid carcinoma  Jiangqiao 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 | |
| |  |  | | --- | --- | | |  | | --- | | [**» Abstract**](https://urldefense.proofpoint.com/v2/url?u=http-3A__alerts.springer.com_re-3Fl-3DD0In67cwjI6gyt153I1e&d=DwMFaQ&c=ZQs-KZ8oxEw0p81sqgiaRA&r=rOMURTZ4fslq9dKzVxltN4xNU-D2Ru1wiL30akyNmwk&m=nj0nvs80NSedvgnUVjyqeFA-89KOaGWZHHZ_lYdP2B0&s=Gc47LSEI_u-BQyWIl7tUaUVY48CCSClsUT3JmETaQ3U&e=)   [**» Full text PDF**](https://urldefense.proofpoint.com/v2/url?u=http-3A__alerts.springer.com_re-3Fl-3DD0In67cwjI6gyt153I1f&d=DwMFaQ&c=ZQs-KZ8oxEw0p81sqgiaRA&r=rOMURTZ4fslq9dKzVxltN4xNU-D2Ru1wiL30akyNmwk&m=nj0nvs80NSedvgnUVjyqeFA-89KOaGWZHHZ_lYdP2B0&s=YkADXNmfo8PQF1EW_gu5qa_Mtj60YJTIN8TG5IrlGKg&e=) | | |
| |  |  |  | | --- | --- | --- | | |  |  | | --- | --- | | Research Paper |  |   Whole-exome sequencing identified compound heterozygous variants in*MMKS* in a Chinese pedigree with Bardet-Biedl syndrome  Zhan Qi, Ying Shen, Qian Fu, Wei Li, Wei Yang, Wenshan Xu, Ping Chu, Yaxin Zhang & Hui Wang | |
| |  |  | | --- | --- | | |  | | --- | | [**» Abstract**](https://urldefense.proofpoint.com/v2/url?u=http-3A__alerts.springer.com_re-3Fl-3DD0In67cwjI6gyt153I1g&d=DwMFaQ&c=ZQs-KZ8oxEw0p81sqgiaRA&r=rOMURTZ4fslq9dKzVxltN4xNU-D2Ru1wiL30akyNmwk&m=nj0nvs80NSedvgnUVjyqeFA-89KOaGWZHHZ_lYdP2B0&s=3TWt6KWU8vr1pqxnn0x1-Ev7-i2XuF4-mlYBXOlZUxM&e=)   [**» Full text PDF**](https://urldefense.proofpoint.com/v2/url?u=http-3A__alerts.springer.com_re-3Fl-3DD0In67cwjI6gyt153I1h&d=DwMFaQ&c=ZQs-KZ8oxEw0p81sqgiaRA&r=rOMURTZ4fslq9dKzVxltN4xNU-D2Ru1wiL30akyNmwk&m=nj0nvs80NSedvgnUVjyqeFA-89KOaGWZHHZ_lYdP2B0&s=wm8vXtNKsBolasQrndd155-CGGbNTXNzIOa54aBSmE4&e=) | | |
| |  |  |  | | --- | --- | --- | | |  |  | | --- | --- | | Research Paper |  |   The clinical and genetic characteristics in children with mitochondrial disease in China  Fang 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 | |
| |  |  | | --- | --- | | |  | | --- | | [**» Abstract**](https://urldefense.proofpoint.com/v2/url?u=http-3A__alerts.springer.com_re-3Fl-3DD0In67cwjI6gyt153I1i&d=DwMFaQ&c=ZQs-KZ8oxEw0p81sqgiaRA&r=rOMURTZ4fslq9dKzVxltN4xNU-D2Ru1wiL30akyNmwk&m=nj0nvs80NSedvgnUVjyqeFA-89KOaGWZHHZ_lYdP2B0&s=4pL3zTp3dOA3J1uZQuVlZYyYbQMePiI5Arj0Q5m39S4&e=)   [**» Full text PDF**](https://urldefense.proofpoint.com/v2/url?u=http-3A__alerts.springer.com_re-3Fl-3DD0In67cwjI6gyt153I1j&d=DwMFaQ&c=ZQs-KZ8oxEw0p81sqgiaRA&r=rOMURTZ4fslq9dKzVxltN4xNU-D2Ru1wiL30akyNmwk&m=nj0nvs80NSedvgnUVjyqeFA-89KOaGWZHHZ_lYdP2B0&s=Icy88ro11ljSzU1KI-lpF3YJvuVB4Wqs_GCR7ag7NSg&e=) | | |
| |  |  |  | | --- | --- | --- | | |  |  | | --- | --- | | Research Paper |  |   Gene mutations and clinical phenotypes in Chinese children with Blau syndrome  Caifeng Li, Junmei Zhang, Shipeng Li, Tongxin Han, Weiying Kuang, Yifang Zhou, Jianghong Deng & Xiaohua Tan | |
| |  |  | | --- | --- | | |  | | --- | | [**» Abstract**](https://urldefense.proofpoint.com/v2/url?u=http-3A__alerts.springer.com_re-3Fl-3DD0In67cwjI6gyt153I1k&d=DwMFaQ&c=ZQs-KZ8oxEw0p81sqgiaRA&r=rOMURTZ4fslq9dKzVxltN4xNU-D2Ru1wiL30akyNmwk&m=nj0nvs80NSedvgnUVjyqeFA-89KOaGWZHHZ_lYdP2B0&s=LFR4A-vwPfMDx6AX0mQC66PsL6Th-qx6OtIZOU5aEv4&e=)   [**» Full text PDF**](https://urldefense.proofpoint.com/v2/url?u=http-3A__alerts.springer.com_re-3Fl-3DD0In67cwjI6gyt153I1l&d=DwMFaQ&c=ZQs-KZ8oxEw0p81sqgiaRA&r=rOMURTZ4fslq9dKzVxltN4xNU-D2Ru1wiL30akyNmwk&m=nj0nvs80NSedvgnUVjyqeFA-89KOaGWZHHZ_lYdP2B0&s=KQkYHKaj9Dxe9HQlgKCDZeLb7RhGp5gH3G6m7LtxpPg&e=) | | |
| |  |  |  | | --- | --- | --- | | |  |  | | --- | --- | | Research Paper |  |   Analysis of genotypes and phenotypes in Chinese children with tuberous sclerosis complex  Dayong Bai, Junyang Zhao, Li Li, Jun Gao & Xu Wang | |
| |  |  | | --- | --- | | |  | | --- | | [**» Abstract**](https://urldefense.proofpoint.com/v2/url?u=http-3A__alerts.springer.com_re-3Fl-3DD0In67cwjI6gyt153I1m&d=DwMFaQ&c=ZQs-KZ8oxEw0p81sqgiaRA&r=rOMURTZ4fslq9dKzVxltN4xNU-D2Ru1wiL30akyNmwk&m=nj0nvs80NSedvgnUVjyqeFA-89KOaGWZHHZ_lYdP2B0&s=QfJt6BQiWyuaiCM762EwWTwZvsNXMPHM77XPzlZtCAQ&e=)   [**» Full text PDF**](https://urldefense.proofpoint.com/v2/url?u=http-3A__alerts.springer.com_re-3Fl-3DD0In67cwjI6gyt153I1n&d=DwMFaQ&c=ZQs-KZ8oxEw0p81sqgiaRA&r=rOMURTZ4fslq9dKzVxltN4xNU-D2Ru1wiL30akyNmwk&m=nj0nvs80NSedvgnUVjyqeFA-89KOaGWZHHZ_lYdP2B0&s=52CXIg_JLNNB79IZ3qnZ7X85PzeNOtCO6YoLiFBeQBo&e=) | | |
| |  |  |  | | --- | --- | --- | | |  |  | | --- | --- | | Letter to the Editor |  |   Berry syndrome: a rare cardiac malformation with extra-cardiac findings  Jingya 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 | |
| |  |  | | --- | --- | | |  | | --- | | [**» Abstract**](https://urldefense.proofpoint.com/v2/url?u=http-3A__alerts.springer.com_re-3Fl-3DD0In67cwjI6gyt153I1o&d=DwMFaQ&c=ZQs-KZ8oxEw0p81sqgiaRA&r=rOMURTZ4fslq9dKzVxltN4xNU-D2Ru1wiL30akyNmwk&m=nj0nvs80NSedvgnUVjyqeFA-89KOaGWZHHZ_lYdP2B0&s=xsfF-NWwwIaXizYq_ubyx9Rmkc0nsnWxHNmWRihKRhc&e=)   [**» Full text PDF**](https://urldefense.proofpoint.com/v2/url?u=http-3A__alerts.springer.com_re-3Fl-3DD0In67cwjI6gyt153I1p&d=DwMFaQ&c=ZQs-KZ8oxEw0p81sqgiaRA&r=rOMURTZ4fslq9dKzVxltN4xNU-D2Ru1wiL30akyNmwk&m=nj0nvs80NSedvgnUVjyqeFA-89KOaGWZHHZ_lYdP2B0&s=HilAsfo43qmL7_dzB5eCy9KYClc-IC7hK59JQ98hrMQ&e=) | | |
| |  |  |  | | --- | --- | --- | | |  |  | | --- | --- | | Letter to the Editor |  |   Detection of mycobacterial and viral DNA in Kikuchi-Fujimoto disease: an analysis of 153 Chinese pediatric cases  Zhe Xu, Ying Liu, Haijing Li, Shufang Meng, Alan S. Boyd, Charles W. Stratton, Lin Ma & Yi-Wei Tang | |
| |  |  | | --- | --- | | |  | | --- | | [**» Abstract**](https://urldefense.proofpoint.com/v2/url?u=http-3A__alerts.springer.com_re-3Fl-3DD0In67cwjI6gyt153I1q&d=DwMFaQ&c=ZQs-KZ8oxEw0p81sqgiaRA&r=rOMURTZ4fslq9dKzVxltN4xNU-D2Ru1wiL30akyNmwk&m=nj0nvs80NSedvgnUVjyqeFA-89KOaGWZHHZ_lYdP2B0&s=aWvT3w3yLc0WbL9DDP-BzSC4Q25e43biG4Jt53GnOFk&e=)   [**» Full text PDF**](https://urldefense.proofpoint.com/v2/url?u=http-3A__alerts.springer.com_re-3Fl-3DD0In67cwjI6gyt153I1r&d=DwMFaQ&c=ZQs-KZ8oxEw0p81sqgiaRA&r=rOMURTZ4fslq9dKzVxltN4xNU-D2Ru1wiL30akyNmwk&m=nj0nvs80NSedvgnUVjyqeFA-89KOaGWZHHZ_lYdP2B0&s=bAYumnpcD_2cBC7Fd9rcvld2P_wMti-mvsNVJbWJLac&e=) | | |
| |  |  |  | | --- | --- | --- | | |  |  | | --- | --- | | Letter to the Editor |  |   Analysis of diagnosis and treatment of lipoblastomatosis  Zhiqiang Mo, Xianghui Xie, Huanmin Wang, Hong Qin, Wei Han & Xiaosong Li | |
| |  |  | | --- | --- | | |  | | --- | | [**» Abstract**](https://urldefense.proofpoint.com/v2/url?u=http-3A__alerts.springer.com_re-3Fl-3DD0In67cwjI6gyt153I1s&d=DwMFaQ&c=ZQs-KZ8oxEw0p81sqgiaRA&r=rOMURTZ4fslq9dKzVxltN4xNU-D2Ru1wiL30akyNmwk&m=nj0nvs80NSedvgnUVjyqeFA-89KOaGWZHHZ_lYdP2B0&s=Ki9X4cJuVdkbXj9lWx__HwjF2CEwxuc0viTJDwuE11U&e=)   [**» Full text PDF**](https://urldefense.proofpoint.com/v2/url?u=http-3A__alerts.springer.com_re-3Fl-3DD0In67cwjI6gyt153I1t&d=DwMFaQ&c=ZQs-KZ8oxEw0p81sqgiaRA&r=rOMURTZ4fslq9dKzVxltN4xNU-D2Ru1wiL30akyNmwk&m=nj0nvs80NSedvgnUVjyqeFA-89KOaGWZHHZ_lYdP2B0&s=G75GN-jvgYgYLnmojqHvmDg-XUkcRJOvMlX9eSMyAJo&e=) | | |