OPINION REVIEW

7187  Effects of glucocorticoids on leukocytes: Genomic and non-genomic mechanisms

Jia WY, Zhang JJ

MINIREVIEWS

7195  Apheresis: A cell-based therapeutic tool for the inflammatory bowel disease

Yasmin F, Najeeb H, Naeem U, Moeed A, Koritala T, Surani S

7209  Helicobacter pylori infection and small intestinal bacterial overgrowth—more than what meets the eye

Dharan M, Wozny D

7215  Anatomy of the anterolateral ligament of the knee joint

Park JG, Han SB, Rhim HC, Jeon OH, Jang KM

ORIGINAL ARTICLE

Clinical and Translational Research

7224  Molecular mechanisms of Biyu decoction as treatment for psoriasis: A network pharmacology and molecular docking study

Wang Z, Zhang HM, Guo YR, Li LL

7242  Expression of hepatocyte nuclear factor 4 alpha, wingless-related integration site, and β-catenin in clinical gastric cancer

Hu Q, Li LL, Peng Z, Yi P

Case Control Study

7256  Improved Pittsburgh Sleep Quality Index scores on first postoperative night achieved by propofol anesthesia in patients undergoing ambulatory gynecologic surgery

Hu CH, Chou WY

7265  Efficacy of Guhong injection versus Butylphthalide injection for mild ischemic stroke: A multicenter controlled study

Zhang WW, Xin J, Zhang GY, Zhai QJ, Zhang HM, Wu CS

Retrospective Study

7275  Clinical values of Barcelona Clinic Liver Cancer subgroup and up-to-7 criteria in intermediate stage hepatocellular carcinoma with transcatheter arterial chemoembolization

Lee SW, Peng YC, Lien HC, Ko CW, Tung CF, Chang CS

7285  Intervention effect of encouraging mental and programmed nursing of patients in interventional operating room on their compliance and bad moods

Chi RB, Cai YY, Mao HP
## Contents

**Thrice Monthly Volume 10 Number 21 July 26, 2022**

<table>
<thead>
<tr>
<th>Page</th>
<th>Title</th>
<th>Authors</th>
</tr>
</thead>
<tbody>
<tr>
<td>7293</td>
<td>Preoperative neoadjuvant chemotherapy in patients with breast cancer evaluated using strain ultrasonic elastography</td>
<td>Pan HY, Zhang Q, Wu WJ, Li X</td>
</tr>
<tr>
<td>7302</td>
<td>Risk factors for delayed intracranial hemorrhage secondary to ventriculoperitoneal shunt: A retrospective study</td>
<td>Chen JC, Duan SX, Xue ZB, Yang SY, Li Y, Lai RL, Tan DH</td>
</tr>
<tr>
<td>7314</td>
<td>Sequential treatment of severe pneumonia with respiratory failure and its influence on respiratory mechanical parameters and hemodynamics</td>
<td>Niu BY, Wang G, Li B, Zhen GS, Weng YB</td>
</tr>
<tr>
<td>7324</td>
<td>Effects of alendronate sodium combined with InterTan on osteoporotic femoral intertrochanteric fractures and fracture recurrence</td>
<td>Wang KM, Wei SP, Yin XY, Meng QJ, Kong YM</td>
</tr>
<tr>
<td>7333</td>
<td>Correlation of magnetic resonance imaging quantitative parameters and apparent diffusion coefficient value with pathological breast cancer</td>
<td>Wang Z, Ren GY, Yin Q, Wang Q</td>
</tr>
<tr>
<td>7341</td>
<td>Risk factors for delirium after surgery for craniocerebral injury in the neurosurgical intensive care unit</td>
<td>Chen RY, Zhong CH, Chen W, Lin M, Feng CF, Chen CN</td>
</tr>
</tbody>
</table>

**Observational Study**

<table>
<thead>
<tr>
<th>Page</th>
<th>Title</th>
<th>Authors</th>
</tr>
</thead>
<tbody>
<tr>
<td>7348</td>
<td>Effect of osteoarthritic knee flexion deformity correction by total knee arthroplasty on sagittal spinopelvic alignment in Indian population</td>
<td>Puthiyapura LK, Jain M, Tripathy SK, Puliappadamb HM</td>
</tr>
</tbody>
</table>

**Randomized Controlled Trial**

<table>
<thead>
<tr>
<th>Page</th>
<th>Title</th>
<th>Authors</th>
</tr>
</thead>
<tbody>
<tr>
<td>7365</td>
<td>Comparison of involved-field intensity-modulated radiotherapy combined with S-1 vs radiotherapy alone for elderly patients with esophageal cancer</td>
<td>Liu LH, Yan MH, Di YP, Fu ZG, Zhang XD, Li HQ</td>
</tr>
</tbody>
</table>

**Randomized Clinical Trial**

<table>
<thead>
<tr>
<th>Page</th>
<th>Title</th>
<th>Authors</th>
</tr>
</thead>
</table>

**META-ANALYSIS**

<table>
<thead>
<tr>
<th>Page</th>
<th>Title</th>
<th>Authors</th>
</tr>
</thead>
<tbody>
<tr>
<td>7386</td>
<td>Impact of cancer on mortality rates in patients with sepsis: A meta-analysis and meta-regression of current studies</td>
<td>Xiang MJ, Chen GL</td>
</tr>
</tbody>
</table>
CASE REPORT

7397  Updated clinical and glycomic features of mannosyl-oligosaccharide glucosidase deficiency: Two case reports  

7409  Solitary necrotic nodules of the liver with "ring"-like calcification: A case report  
   *Bao JP, Tian H, Wang HC, Wang CC, Li B*

7415  Corticosteroid-induced bradycardia in multiple sclerosis and maturity-onset diabetes of the young due to hepatocyte nuclear factor 4-alpha mutation: A case report  
   *Sohn SY, Kim SY, Joo IS*

7422  Essential thrombocythemia with non-ST-segment elevation myocardial infarction as the first manifestation: A case report  
   *Wang ZM, Chen WH, Wu YM, Wang LQ, Ye FL, Yin RL*

7429  Extranasopharyngeal angiofibroma in children: A case report  
   *Yan YY, Lai C, Wu L, Fu Y*

7438  Deep Sylvian fissure meningiomas: A case report  

7445  Acute pulmonary embolism originating from upper limb venous thrombosis following breast cancer surgery: Two case reports  
   *Duan Y, Wang GL, Guo X, Yang LL, Tian FG*

7451  Managing spondylitis tuberculosis in a patient with underlying diabetes and hypothyroidism: A case report  
   *Novita BD, Muliono AC, Wijaya S, Theodora I, Tjahjono Y, Supit VD, Willianto VM*

7459  Ovarian mucinous tumor with mural nodules of anaplastic carcinoma: Three case reports  
   *Wang XJ, Wang CY, Xi YF, Bu P, Wang P*

7467  Transcatheter arterial infusion chemotherapy and embolization for primary lacrimal sac squamous cell carcinoma: A case report  
   *Sun MH, Yi WD, Shen L, Zhou L, Lu JX*

7474  Programmed cell death-1 inhibitor combination treatment for recurrent proficient mismatch repair/microsatellite-stable type endometrial cancer: A case report  
   *Zhai CY, Yin LX, Han WD*

7483  Novel compound heterozygous mutation of SLC12A3 in Gitelman syndrome co-existent with hyperthyroidism: A case report and literature review  
   *Qin YZ, Liu YM, Wang Y, You C, Li LN, Zhou XY, Lv WM, Hong SH, Xiao LX*

7495  Successful treatment of hyperglycemia with liraglutide in a hospitalized 27-year-old patient with schizophrenia: A case report  
   *Zhang L, Yu WJ, Zhu H, Li HF, Qiao J*
<table>
<thead>
<tr>
<th>Page</th>
<th>Title</th>
<th>Authors</th>
</tr>
</thead>
<tbody>
<tr>
<td>7502</td>
<td>Refractory lymphoma treated with chimeric antigen receptor T cells combined with programmed cell death-1 inhibitor: A case report</td>
<td>Zhang CJ, Zhang JY, Li LJ, Xu NW</td>
</tr>
<tr>
<td>7509</td>
<td>Median arcuate ligament syndrome with retroperitoneal haemorrhage: A case report</td>
<td>Lu XC, Pei JG, Xie GH, Li YY, Han HM</td>
</tr>
<tr>
<td>7517</td>
<td>Novel frameshift mutation in the AHDC1 gene in a Chinese global developmental delay patient: A case report</td>
<td>Lin SZ, Xie HY, Qu YL, Gao W, Wang WQ, Li JY, Feng XC, Jin CQ</td>
</tr>
<tr>
<td>7523</td>
<td>Selective nerve block for the treatment of neuralgia in Kummell’s disease: A case report</td>
<td>Zhang X, Li ZX, Yin LJ, Chen H</td>
</tr>
<tr>
<td>7531</td>
<td>Traditional Chinese medicine manipulative reduction combined with percutaneous vertebroplasty for treating type III Kummell’s disease: A case report</td>
<td>Hao SS, Zhang RJ, Dong SL, Li HK, Liu S, Li RF, Ren HH, Zhang LY</td>
</tr>
<tr>
<td>7539</td>
<td>Differential diagnosis and treatment of foot drop caused by an extraneural ganglion cyst above the knee: A case report</td>
<td>Won KH, Kang EY</td>
</tr>
<tr>
<td>7553</td>
<td>Chronic urticaria associated with lung adenocarcinoma — a paraneoplastic manifestation: A case report and literature review</td>
<td>Jiménez LF, Castellón EA, Marenco JD, Mejía JM, Rojas CA, Jiménez FT, Coronell L, Osorio-Llanes E, Mendoza-Torres E</td>
</tr>
<tr>
<td>7565</td>
<td>Spinal giant cell-rich osteosarcoma-diagnostic dilemma and treatment strategy: A case report</td>
<td>Tseng CS, Wong CE, Huang CC, Hsu HH, Lee JS, Lee PH</td>
</tr>
<tr>
<td>7571</td>
<td>Primary clear cell sarcoma of soft tissue in the posterior cervical spine invading the medulla oblongata: A case report</td>
<td>Liu CC, Huang WP, Gao JB</td>
</tr>
<tr>
<td>7577</td>
<td>Pseudomonas aeruginosa-related effusive-constrictive pericarditis diagnosed with echocardiography: A case report</td>
<td>Chen JI, Mei DE, Yu CG, Zhao ZY</td>
</tr>
<tr>
<td>7592</td>
<td>Considerations of single-lung ventilation in neonatal thoracoscopic surgery with cardiac arrest caused by bilateral pneumothorax: A case report</td>
<td>Zhang X, Song HC, Wang KL, Ren YY</td>
</tr>
<tr>
<td>Page</td>
<td>Title</td>
<td>Authors</td>
</tr>
<tr>
<td>------</td>
<td>----------------------------------------------------------------------</td>
<td>-------------------------------------------------------------------------</td>
</tr>
<tr>
<td>7599</td>
<td>Rare primary rectal mucosa-associated lymphoid tissue lymphoma with curative resection by endoscopic submucosal dissection: A case report and review of literature</td>
<td>Tao Y, Nan Q, Lei Z, MiaoYL, Niu JK</td>
</tr>
<tr>
<td>7609</td>
<td>Differences in examination results of small anastomotic fistula after radical gastrectomy with afterward treatments: A case report</td>
<td>Lu CY, LiuYL, LiuKJ, Xu S, Yao HL, Li L, Guo ZS</td>
</tr>
<tr>
<td>7617</td>
<td>Baseline differences may impact on relationship between dietary tryptophan and risk of obesity and type 2 diabetes</td>
<td>Ren XH, Ye YW, He LP</td>
</tr>
</tbody>
</table>
ABOUT COVER
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WJCC mainly publishes articles reporting research results and findings obtained in the field of clinical medicine and covering a wide range of topics, including case control studies, retrospective cohort studies, retrospective studies, clinical trials studies, observational studies, prospective studies, randomized controlled trials, randomized clinical trials, systematic reviews, meta-analysis, and case reports.

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RESPONSIBLE EDITORS FOR THIS ISSUE
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Novel frameshift mutation in the \textit{AHDC1} gene in a Chinese global developmental delay patient: A case report

Shuang-Zhu Lin, Hong-Yan Xie, Yan-Lai Qu, Wen Gao, Wan-Qi Wang, Jia-Yi Li, Xiao-Chun Feng, Chun-Quan Jin

\textbf{Abstract}

\textbf{BACKGROUND}  
Xia–Gibbs syndrome (XGS, OMIM: 615829), caused by mutations within the AT-Hook DNA-binding motif-containing protein 1 (\textit{AHDC1}) gene (OMIM: 615790), located on the short arm of chromosome 1 within the cytogenetic band 1p36.11, contains five noncoding 5 exons, a single 4.9-kb coding exon, and a noncoding 3 exon.

\textbf{CASE SUMMARY}  
In this case report, we diagnosed and treated a 6-mo-old girl with XGS. The primary clinical symptoms included global developmental delay, hypotonia, and mild dysmorphic features. Using high-throughput whole-exome sequencing to sequence the patient and her parents, and the results showed a novel frameshift mutation of c.1155dupG (p.Arg386Alafs*3) in the \textit{AHDC1} gene. The paternal gene was wild type.

\textbf{CONCLUSION}  
This report extends the mutation spectrum of the \textit{AHDC1} gene to provide the diagnostic basis for genetic counseling in families with XGS.

\textbf{Key Words:} Xia–Gibbs syndrome; AT-Hook DNA-binding motif-containing protein 1; Children; Global developmental delay; Case report

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Core Tip: We report a 6-mo-old girl with Xia–Gibbs syndrome (XGS). The main clinical manifestations were global developmental delay, hypotonia, and other mild dysmorphic features. DNA sequencing showed that there was a novel frameshift mutation of c.1155dupG (p.Arg386Alafs*3) in the AT-Hook DNA-binding motif-containing protein 1 (AHDC1) gene. This study extends the mutation spectrum of the AHDC1 gene, and provides a molecular basis for the etiological diagnosis of XGS and genetic consultation for the family.

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INTRODUCTION
Xia–Gibbs syndrome (XGS) is an autosomal dominant genetic disease caused by mutation of the AT-Hook DNA-binding motif-containing protein 1 (AHDC1) gene. Typical features include global developmental delay, hypotonia, obstructive sleep apnea, seizures, delayed myelination, micrognathia, and other mild dysmorphic features[1,2]. The AHDC1 gene is located on the short arm of chromosome 1 within the cytogenetic band 1p36.11, and consists of seven exons with only one coding exon (exon 6), five noncoding 5 exons, and a noncoding 3 exon[3].

In this case, a novel frameshift mutation of c.1155dupG (p.Arg386Alafs*3) in the AHDC1 gene was found by high-throughput whole-exome sequencing (WES) in a patient with global developmental delay, hypotonia, micrognathia, and other mild dysmorphic features.

CASE PRESENTATION
Chief complaints
A 6-mo-old female patient presented to our hospital due to global developmental delay.

History of present illness
In June 2021, the patient presented with a 6-mo history of global developmental delay. Since birth, the child had lagged behind in developmental milestones. She could not raise her head until she was age 4 mo, and she still shook her head sometimes. She could not sit alone without the help of others, and turning over was not flexible. She could not crawl, and the stability of her lower limbs was too poor to support her weight. She had no difficulty in pronunciation.

History of past illness
The patient weighed 3.7 kg and was 52 cm long after full-term normal delivery without a history of asphyxiation and resuscitation. The patient did not have a history of feeding difficulties. There was no history of encephalitis and brain trauma since disease onset.

Personal and family history
Her parents were clinically normal. Genetic history or family history of infectious diseases were denied.

Physical examination
At her visit at 6 mo of age, we performed a physical examination. Her weight was 9.0 kg and length 70.0 cm. Mild dysmorphic features were observed, such as micrognathia. Grasping reflex was present, and limb muscle tension showed hypotonia. Ankle clonus was positive. Muscle strength was level 4. Physiological reflexes were present.

Laboratory examinations
We performed a series of examinations of the patient, including liver function, kidney function, electrolytes, and organic acids in blood and urine, which showed no abnormalities.

Imaging examinations
In June 2021, brain magnetic resonance imaging showed bilateral frontal subdural effusion, hydrocephalus, and hydrops in the mastoid area. Electroencephalography (EEG) was abnormal,
showing sharp waves and spike waves in the Rolandic area or right forehead region.

**Further diagnostic work-up**

The patient underwent a Peabody Developmental Motor Scales (PDMS-2) test on May 14, 2021[4]. The Fine Motor Quotient (FMQ) was 9 points, with a motor quotient of 67, and the Gross Motor Quotient (GMQ) was 21 points with a motor quotient of 81; both measures were worse than those of her peers. Griffiths Mental Development Scales (GMDS) showed that motor ability was equivalent to that of a 3-mo-old child, with a development quotient (DQ) of 46 points; human-social ability was equivalent to that of a 4-mo-old child, with a DQ of 62 points; hearing and language ability was equivalent to that of a 4.5-mo-old child with a DQ of 69 points, and hand and eye coordination was equivalent to that of a 3.5-mo-old child, with a DQ of 54 points. The patients’ overall performance was equivalent to that of a 3.5-mo-old child, with a DQ of 54 points.

**High throughput WES and mitochondrial sequencing**

Informed parental consent was obtained for WES, mitochondrial sequencing, and publication of photographs on behalf of the proband. DNA samples were extracted from the peripheral blood of the child and her parents to detect whole-exome sequences and whole-genome copy number variants. The results revealed a novel frameshift mutation of c.1155dupG (p.Arg386Alafs*3) in the *AHDC1* gene. Polymorphic sites were detected in each sample data using GATK software and statistical analysis was performed on 1000 human genomes and ExAC databases. The reported pathogenic locus has been confirmed using the Human Gene Mutation Database (HGMD) and the Human Online Mendelian Genetic Database (OMIM). The pathogenicity of the mutation locus was comprehensively assessed using the American College of Medical Genetics and Genomics (ACMG) standards and guidelines for the interpretation of sequence variation.

**Gene detection and pathogenicity analysis**

The WES results showed the presence of a novel frameshift variant in the *AHDC1* gene, which was an unreported frameshift mutation, c.1155dupG (p.Arg386Alafs*3), and may result in altered gene function. The frequency at which variation occurs in the normal population database is unknown, and is a low-frequency variation. The results of protein function prediction are unknown, and are not reported in the HGMD database.

According to Sanger sequencing, the variation occurred in the child, while the parental genes were wild type (Figures 1-3). The mutation was suspected to be a pathogenic according to the ACMG guidelines[5].

**FINAL DIAGNOSIS**

Sanger sequencing showed that there was a novel frameshift variation of c.1155dupG (p.Arg386Alafs*3) in the *AHDC1* gene. Based on clinical presentation, laboratory tests and gene sequencing results, the clinical diagnosis was XGS.

**TREATMENT**

To improve quality of life of the patient, she has been provided with rehabilitation training and behavioral guidance therapy since June 2021.

**OUTCOME AND FOLLOW-UP**

The patient had been receiving rehabilitation treatment for nearly 6 mo. The patient is now age 11 mo, her weight is 11.0 kg and length is 74.0 cm. She can sit alone, and her hands can engage in more elaborate movements, and have increased responses to external stimuli, and she is able to speak some simple vowels. The patient underwent the PDMS-2 test in November 2021. The FMQ was 13 points with a motor quotient of 79, and GMQ was 17 points with a motor quotient of 72. GMDS showed that motor ability was equivalent to that of a 5.3-mo-old child, with a DQ of 46 points. Performance was equivalent to that of a 6.5-mo-old child, with a DQ of 56 points.
Lin SZ et al. Novel frameshift mutation in AHDC1

Figure 1 A novel frameshift variation in c.1155dupG (p.Arg386Alafs*3) of the AT-Hook DNA-binding motif-containing protein 1 gene was revealed by Sanger sequencing.

Figure 2 No anomalies were found in the AT-Hook DNA-binding motif-containing protein 1 gene in the child's father as revealed by Sanger sequencing.

Figure 3 No anomalies were found in the AT-Hook DNA-binding motif-containing protein 1 gene in the child's mother as revealed by Sanger sequencing.

DISCUSSION

The AHDC1 gene is located on chromosome 1p36.11, and likely functions in DNA binding. The AHDC1 gene is part of the CBX family of proteins associated with human chromodomain-containing Polycomb proteins. It encodes a protein of 1603 amino acids, consisting of five noncoding 5 exons, a noncoding 3 exon and a single 4.9-kb coding exon (exon 6) containing 2 AT hooks[1,3]. Previous studies have shown that AHDC1 interacts with nuclear proteins involved in epigenetic regulation during development, mainly at neural loci and neuronal protein transport. Mutation of the AHDC1 gene can lead to XGS.

XGS (OMIM: 615829) is an autosomal dominant genetic disease caused by mutation of the AHDC1 gene. Typical features include global developmental delay, intellectual disability, structural abnormalities of the brain, global hypotonia, feeding problems, sleep difficulties, apnea, and short stature[6]. In addition to neurological seizures, delayed myelination, micrognathia, and other mild dysmorphic manifestations[7], XGS patients may also have a broad clinical range and multisystem involvement[8,9]. Not all XGS patients show a typical phenotype, and there are differences in the disease manifestations [10,11].

The present patient first attended hospital due to delayed motor milestones over 2 mo when she was age 6 mo. In order to clarify the cause of the disease, we used high-throughput WES and identified an unreported mutation of the AHDC1 gene.
According to previous studies, more than 90% of patients had motor and speech delay, hypotonia occurred in approximately 85% of patients, and less than 40% of patients had short stature. About 30% of patients had symptoms of autism. Some patients showed sleep apnea (34.33%), laryngomalacia (14.93%), and other manifestations. Approximately 35% of patients developed epileptic seizures[8,12,13].

In this case report, the child conformed to the typical clinical manifestations of XGS. The child developed significant motor delay and hypotonia, but speech ability was not delayed. She was able to respond positively to external stimuli. There was no sufficient evidence of sleep apnea and laryngomalacia. Although the child’s EEG showed the distribution of spike waves and sharp waves, she did not appear to have the related actions, and she did not have other related manifestations of epilepsy [14,15].

Our patient has not yet developed growth hormone deficiency or short stature. Height and weight were within the mean range of normal, in contrast to two previously reported Chinese children who exhibited partial growth hormone deficiency and attended hospital due to short stature[16].

In this report, we describe a Chinese patient with XGS, with a new mutation c.1155dupG (p.Arg386Alafs*3) in the AHDC1 gene identified by WES, and compared the results with those from two Chinese cases reported in the literature, to better understand the clinical phenotype and the association with the AHDC1 gene.

CONCLUSION

Previous studies have shown that global developmental delay occurs in the AHDC1-related phenotype of XGS. Our patient was found to have a novel frameshift variation of c.1155dupG (p.Arg386Alafs*3) in the AHDC1 gene, which was an unreported frameshift mutation. Typical features of XGS include global developmental delay, hypotonia, obstructive sleep apnea, seizures, delayed myelination, micrognathia, and other mild dysmorphic features. This report result showed clinicians could consider XGS in patients with similar clinical characteristics. Genetic testing can help physicians confirm the diagnosis and help with further genetic counseling.

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We would like to thank the family members of the child for agreeing to participate in this study.

FOOTNOTES

Author contributions: Lin SZ and Xie HY collected and analyzed all clinical data and wrote the manuscript; Qu YL and Gao W participated in the collection of the literature and the chart research; Jin CQ was involved in the genetic diagnosis and treatment of the patients; Lin SZ, Wang WQ, Li JY and Feng XC substantially participated in drafting and revising the important intellectual content of the manuscript; All authors involved have read and approved the final manuscript.

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REFERENCES


