## OPINION REVIEW

**Malignant insulinoma: Can we predict the long-term outcomes?**

*Cigrovski Berkovic M, Uglamec M, Marinovic S, Balen I, Mrzljak A*

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## MINIREVIEWS

**Practical points that gastrointestinal fellows should know in management of COVID-19**

*Sahin T, Simsek C, Balaban HY*

**Nanotechnology in diagnosis and therapy of gastrointestinal cancer**

*Liang M, Li LD, Li L, Li S*

**Advances in the clinical application of oxycodone in the perioperative period**

*Chen HY, Wang ZN, Zhang WY, Zhu T*

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## ORIGINAL ARTICLE

### Clinical and Translational Research

**Circulating miR-627-5p and miR-199a-5p are promising diagnostic biomarkers of colorectal neoplasia**

*Zhao DY, Zhou L, Yin TF, Zhou YC, Zhou GYJ, Wang QQ, Yao SK*

### Retrospective Cohort Study

**Management and outcome of bronchial trauma due to blunt versus penetrating injuries**

*Gao JM, Li H, Du DY, Yang J, Kong LW, Wang JB, He P, Wei GB*

### Retrospective Study

**Ovarian teratoma related anti-N-methyl-D-aspartate receptor encephalitis: A case series and review of the literature**

*Li SJ, Yu MH, Cheng J, Bai WX, Di W*

**Endoscopic surgery for intraventricular hemorrhage: A comparative study and single center surgical experience**

*Wang FB, Yuan XW, Li JX, Zhang M, Xiang ZH*

**Protective effects of female reproductive factors on gastric signet-ring cell carcinoma**

*Li Y, Zhong YX, Xu Q, Tian YT*

**Risk factors of mortality and severe disability in the patients with cerebrovascular diseases treated with perioperative mechanical ventilation**

*Zhang JZ, Chen H, Wang X, Xu K*
<table>
<thead>
<tr>
<th>Page</th>
<th>Title</th>
<th>Authors</th>
</tr>
</thead>
<tbody>
<tr>
<td>5266</td>
<td>Long-term outcomes of high-risk percutaneous coronary interventions under extracorporeal membrane oxygenation support: An observational study</td>
<td>Huang YX, Xu ZM, Zhao L, Cao Y, Chen Y, Qiu YG, Liu YM, Zhang PY, He JC, Li TC</td>
</tr>
<tr>
<td>5275</td>
<td>Health care worker occupational experiences during the COVID-19 outbreak: A cross-sectional study</td>
<td>Li XF, Zhou XL, Zhao SX, Li YM, Pan SQ</td>
</tr>
<tr>
<td>5287</td>
<td>Enhanced recovery after surgery strategy to shorten perioperative fasting in children undergoing non-gastrointestinal surgery: A prospective study</td>
<td>Ying Y, Xu HZ, Han ML</td>
</tr>
<tr>
<td>5297</td>
<td>Orthodontic treatment combined with 3D printing guide plate implant restoration for edentulism and its influence on mastication and phonic function</td>
<td>Yan LB, Zhou YC, Wang Y, Li LX</td>
</tr>
<tr>
<td>5306</td>
<td>Effectiveness of psychosocial intervention for internalizing behavior problems among children of parents with alcohol dependence: Randomized controlled trial</td>
<td>Omkarappa DB, Rentala S, Natalla P</td>
</tr>
<tr>
<td>5317</td>
<td>Crouzon syndrome in a fraternal twin: A case report and review of the literature</td>
<td>Li XJ, Su JM, Ye XW</td>
</tr>
<tr>
<td>5324</td>
<td>Laparoscopic duodenojejunostomy for malignant stenosis as a part of multimodal therapy: A case report</td>
<td>Murakami T, Matsui Y</td>
</tr>
<tr>
<td>5331</td>
<td>Chordoma of petrosal mastoid region: A case report</td>
<td>Hua JJ, Ying ML, Chen ZW, Huang C, Zheng CS, Wang YJ</td>
</tr>
<tr>
<td>5337</td>
<td>Pneumatosis intestinalis after systemic chemotherapy for colorectal cancer: A case report</td>
<td>Liu H, Hsieh CT, Sun JM</td>
</tr>
<tr>
<td>5343</td>
<td>Mammary-type myofibroblastoma with infarction and atypical mitosis-a potential diagnostic pitfall: A case report</td>
<td>Zeng YF, Dai YZ, Chen M</td>
</tr>
<tr>
<td>Page</td>
<td>Title</td>
<td>Authors</td>
</tr>
<tr>
<td>------</td>
<td>-----------------------------------------------------------------------</td>
<td>------------------------------------------------------------------------</td>
</tr>
<tr>
<td>5365</td>
<td>Neonatal hemorrhage stroke and severe coagulopathy in a late preterm infant after receiving umbilical cord milking: A case report</td>
<td>Lu Y, Zhang ZQ</td>
</tr>
<tr>
<td>5373</td>
<td>Heel pain caused by os subcalcis: A case report</td>
<td>Saijilafu, Li SY, Yu X, Li ZQ, Yang G, Lv JH, Chen GX, Xu RJ</td>
</tr>
<tr>
<td>5380</td>
<td>Pulmonary lymphomatoid granulomatosis in a 4-year-old girl: A case report</td>
<td>Yao JW, Qiu L, Liang P, Liu HM, Chen LN</td>
</tr>
<tr>
<td>5387</td>
<td>Idiopathic membranous nephropathy in children: A case report</td>
<td>Cui KH, Zhang H, Tao YH</td>
</tr>
<tr>
<td>5394</td>
<td>Successful treatment of aortic dissection with pulmonary embolism: A case report</td>
<td>Chen XG, Shi SY, Ye YY, Wang H, Yao WF, Hu L</td>
</tr>
<tr>
<td>5400</td>
<td>Renal papillary necrosis with urinary tract obstruction: A case report</td>
<td>Pan HH, Luo YJ, Zhu QG, Ye LF</td>
</tr>
<tr>
<td>5414</td>
<td>Successful living donor liver transplantation with a graft-to-recipient weight ratio of 0.41 without portal flow modulation: A case report</td>
<td>Kim SH</td>
</tr>
<tr>
<td>5420</td>
<td>Treatment of gastric hepatoid adenocarcinoma with pembrolizumab and bevacizumab combination chemotherapy: A case report</td>
<td>Liu M, Luo C, Xie ZZ, Li X</td>
</tr>
<tr>
<td>5428</td>
<td>Ipsilateral synchronous papillary and clear renal cell carcinoma: A case report and review of literature</td>
<td>Yin J, Zheng M</td>
</tr>
<tr>
<td>5441</td>
<td>PIGN mutation multiple congenital anomalies-hypotonia-seizures syndrome 1: A case report</td>
<td>Hou F, Shan S, Jin H</td>
</tr>
<tr>
<td>Page</td>
<td>Title</td>
<td>Authors</td>
</tr>
<tr>
<td>------</td>
<td>----------------------------------------------------------------------</td>
<td>----------------------------------------------</td>
</tr>
<tr>
<td>5446</td>
<td>Pediatric acute myeloid leukemia patients with i(17)(q10) mimicking acute promyelocytic leukemia: Two case reports</td>
<td>Yan HX, Zhang WH, Wen JQ, Liu YH, Zhang BJ, Ji AD</td>
</tr>
<tr>
<td>5456</td>
<td>Fatal left atrial air embolism as a complication of percutaneous transthoracic lung biopsy: A case report</td>
<td>Li YW, Chen C, Xu Y, Weng QP, Qian SX</td>
</tr>
<tr>
<td>5463</td>
<td>Diagnostic value of bone marrow cell morphology in visceral leishmaniasis-associated hemophagocytic syndrome: Two case reports</td>
<td>Shi SL, Zhao H, Zhou BJ, Ma MB, Li XJ, Xu J, Jiang HC</td>
</tr>
<tr>
<td>5470</td>
<td>Rare case of hepatocellular carcinoma metastasis to urinary bladder: A case report</td>
<td>Kim Y, Kim YS, Yoo JJ, Kim SG, Chin S, Moon A</td>
</tr>
<tr>
<td>5479</td>
<td>Osteotomy combined with the trephine technique for invisible implant fracture: A case report</td>
<td>Chen LW, Wang M, Xia HB, Chen D</td>
</tr>
<tr>
<td>5487</td>
<td>Clinical diagnosis, treatment, and medical identification of specific pulmonary infection in naval pilots: Four case reports</td>
<td>Zeng J, Zhao GL, Yi JC, Liu DD, Jiang YQ, Lu X, Liu YB, Xue F, Dong J</td>
</tr>
<tr>
<td>5502</td>
<td>Mixed large and small cell neuroendocrine carcinoma of the stomach: A case report and review of literature</td>
<td>Li ZF, Lu HZ, Chen YT, Bai XF, Wang TB, Fei H, Zhao DB</td>
</tr>
</tbody>
</table>

**LETTER TO THE EDITOR**

<table>
<thead>
<tr>
<th>Page</th>
<th>Title</th>
<th>Authors</th>
</tr>
</thead>
<tbody>
<tr>
<td>5510</td>
<td>Pleural involvement in cryptococcal infection</td>
<td>Georgakopoulou VE, Damaskos C, Sklapani P, Trakas N, Gkoufa A</td>
</tr>
<tr>
<td>5515</td>
<td>Electroconvulsive therapy plays an irreplaceable role in treatment of major depressive disorder</td>
<td>Ma ML, He LP</td>
</tr>
</tbody>
</table>
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PIGN mutation multiple congenital anomalies-hypotonia-seizures syndrome 1: A case report

Fei Hou, Shan Shan, Hua Jin

**Abstract**

**BACKGROUND**

Multiple congenital anomalies-hypotonia-seizures syndrome 1 (MCAHS1) associated with mutations in *PIGN* gene.

**CASE SUMMARY**

The authors report 1 case of a 16 years old girl who was presented with epilepsy, developmental delay and cerebellar atrophy. She harbors a compound heterozygous variant in the *PIGN* gene, include a nonsense splice site mutation (c.2557A>C) which was inherited from her mother, and a novel site mutation (c.980del) which was inherited from her father.

**CONCLUSION**

This case report expands the mutation spectrum found in *PIGN* gene, and strengthens the association between *PIGN* mutation and MCAHS1. Mutations in *PIGN* gene may be an underestimated cause of epilepsy. The authors recommend that, for patients with epilepsy or prenatal diagnosis of highly suspicious fetus, gene sequencing should be the preferred detection method.

**Key Words:** PIGN; Multiple congenital anomalies-hypotonia-seizures Syndrome 1; MCAHS1; Whole-exome sequencing

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Core Tip: We report 1 case of a 16-year-old girl who presented with epilepsy, developmental delay, and cerebellar atrophy. She harbors compound heterozygous variants in the PIGN gene, including a nonsense splice site mutation (c.2557A>C) that was inherited from her mother and a novel site mutation (c.980del) that was inherited from her father. The maternally inherited variant (c.2557A>C) has not been observed in the gnomAD and 1000genomes, which was called variants of unknown significance. The novel mutation c.980del (paternally inherited) that was detected in the prohand was predicted to be “probably damaging”.

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INTRODUCTION
Multiple congenital anomalies-hypotonia-seizures syndrome 1 (MCAHS1) associated with mutations in PIGN gene, is an autosomal recessive disease featured as epilepsy, stunting, hypotonica, and various congenital disorders. This was initially described in 2011[1]. Mutation in the PIGN gene involved in GPI-anchor pathway have been identified cause varied neurological abnormalities[1-4]. The authors report on a 16 years old girl who was presented with epilepsy, developmental delay and cerebellar atrophy. She harbors a compound heterozygous variant in the PIGN gene, include a nonsense splice site mutation (c.2557A>C) which was inherited from her mother, and a novel site mutation (c.980del) which was inherited from her father.

CASE PRESENTATION

**Chief complaints**
A female, 16 years old, was presented with epilepsy, developmental delay and cerebellar atrophy.

**History of present illness**
Weakness of muscles was noticed at age 2 years old. At 1 year of age, she developed clinical seizures. Her seizure types include twitching movements, starting episodes, cluster seizures, and spasms. At a year and a half, there was only partial response to anti-convulsive therapy. Subsequently, global developmental delay was noted by 2 years old of age, there was no speech, and she needed assistance with daily life activities.

**History of past illness**
She was vigorous in the newborn period and passed her newborn disease screening, began walking around the age of 18 mo.

**Personal and family history**
The patient was the second child of the family (non-consanguineous), her mother was 33 years old. She was born at full term with normal birth parameters, the pregnancy was normal, there was no known teratogenic exposure.

The parents does not have a personal history of seizures, nor is there a family history seizures.

**Physical examination**
Clinical examination documented the eyes were deep set with nystagmus, a small nose with nasalbridge (Figure 1).

**Laboratory examinations**
Whole exome sequencing (WES) was performed commercially at Berrygenomics on the patient and her parents. Sequencing was performed on Illumina Novaseq 6000 (Illumina, San Diego, United States), using the Nano WES Human Exome V1.0 (Berrygenomics, Beijing, China) kit with 200 bp paired-end read, according to the manufacturer’s instructions. Annotation of variants was performed using GATK (https://software.broadinstitute.org/gatk/), gnomAD (http://gnomad.broadinstitute.org/), 1000genomes (http://browser.1000genomes.org), SIFT (http://sift.jcvi.org), FATHMM (http://fathmm.biocompute.org.uk), MutationAssessor (http://mutationassessor.org), OMIM (http://www.omim.org), ClinVar (http://www.ncbi.nlm.nih.gov/clinvar), HGMD (http://
Sanger sequencing

Sequence analysis of exon 28 and 12 of PIGN, using genomic DNA from the patient and her parents, was performed by amplification of a 438 bp and 239 bp fragment containing the putative mutation identified via exome sequencing. The sense primer sequence was 5′-TAAGTCAGTTTCATCAC-GTTCTAT-3′ and the antisense primer sequence was 5′-ATTTCCTCTAATGACAAGCAACAC-3′ for exon 28. The sense primer sequence was 5′-TCTAGCAAATGACACTTTTAGAGA-3′, and the antisense primer sequence was 5′-TTCCITACCCTTACGAGTTAAGAG-3′ for exon 12.

Imaging examinations

Neuropsychological testing demonstrated moderate intellectual disability. Brain imaging showed the progression of mild global cerebral volume loss and cerebellar atrophy. The parents could not provide the results of previous tests, including brain magnetic resonance imaging, various metabolic tests.

FINAL DIAGNOSIS

Whole Exome sequencing was performed commercially at BerryGenomics on the patient and her parents. Compound heterozygous mutations in PIGN gene were found, maternally inherited c.2557A>C and paternally inherited c.980del. The mutations were validated by sanger sequencing in the patient and her parents (Figure 2).

The authors also examined several small insertion/deletion variants (Indels), but none of these had an apparent connection to the clinical phenotype.

TREATMENT

The present therapies for patients with PIGN gene associated illnesses are mainly supportive, which were aimed to reduce the development of epileptic seizures.

OUTCOME AND FOLLOW-UP

The authors will keep on to follow the patient’s disease development. This diagnosis allowed permitted appropriate genetic counseling with related risk evaluation.
DISCUSSION

There are more than 20 genes that participated in GPI-anchor biosynthesis pathway. *PIGN* is responsible for supplement of phosphoethanolamine to the primary mannose in GPI [2]. Mutations in the *PIGN* gene involved in GPI biosynthesis, have been identified associated with MCAHS1 [1,5,6]. The authors report a patient with epilepsy, global delay, cerebellar atrophy with a *PIGN* mutation. The maternally inherited variant (c.2557A>C) has not been seen observed in the gnomAD and 1000genomes, which was called variants of unknown significance. The original mutation c.980del (paternally inherited) discovered in the prohand was estimated as “probably damaging”. The novel mutation changes the gene’s open reading frameshift, support the conclusion that the novel mutation detected in the patient cause major damage to the GPI-anchored protein, finally leading to the disorder. Unlike previous reports [1,5,6], the patient did not have other visceral congenital anomalies of urinary, cardiac or gastrointestinal systems. Gastro-esophageal reflux, diaphragmatic hernia, brachycephaly, flat face, hypoplasia of distal parts of all fingers, open mouth, drooling have not been seen in the patient. Evaluation for mutations in *PIGN* causing *PIGN* associated epilepsy or MCAHS1 should be considered in patients of all ethnicities with epilepsy. These phenotypic differences may be explained as allele specific effects.

The clinical severity of the disease seems to correlate with the predicted functional severity of the mutations seen in *PIGN*[6]. Depending upon the severity of mutations, major congenital anomalies may also be present. Other hypotheses of environmental and genetic modification need to be considered. It should also be noted here that for *PIGN* gene-associated with disability, some tissues are more sensitive than others during body development.

CONCLUSION

This case report expands the mutation spectrum found in *PIGN* gene, and strengthens the association between *PIGN* mutation and MCAHS1. Mutations in *PIGN* gene may be an underestimated cause of epilepsy. The authors recommend that, for patients with epilepsy or prenatal diagnosis of highly suspicious fetus, gene sequencing should be the preferred detection method.

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FOOTNOTES

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