

World Journal of *Clinical Cases*

World J Clin Cases 2021 August 26; 9(24): 6964-7291



OPINION REVIEW

- 6964 Reconsideration of recurrence and metastasis in colorectal cancer
Wang R, Su Q, Yan ZP

MINIREVIEWS

- 6969 Multiple immune function impairments in diabetic patients and their effects on COVID-19
Lu ZH, Yu WL, Sun Y
- 6979 Discontinuation of antiviral therapy in chronic hepatitis B patients
Medas R, Liberal R, Macedo G

ORIGINAL ARTICLE**Case Control Study**

- 6987 Textural differences based on apparent diffusion coefficient maps for discriminating pT3 subclasses of rectal adenocarcinoma
Lu ZH, Xia KJ, Jiang H, Jiang JL, Wu M

Retrospective Cohort Study

- 6999 Cost-effective screening using a two-antibody panel for detecting mismatch repair deficiency in sporadic colorectal cancer
Kim JB, Kim YI, Yoon YS, Kim J, Park SY, Lee JL, Kim CW, Park IJ, Lim SB, Yu CS, Kim JC

Retrospective Study

- 7009 Novel model combining contrast-enhanced ultrasound with serology predicts hepatocellular carcinoma recurrence after hepatectomy
Tu HB, Chen LH, Huang YJ, Feng SY, Lin JL, Zeng YY
- 7022 Influence of volar margin of the lunate fossa fragment fixation on distal radius fracture outcomes: A retrospective series
Meng H, Yan JZ, Wang B, Ma ZB, Kang WB, Liu BG
- 7032 Case series of COVID-19 patients from the Qinghai-Tibetan Plateau Area in China
Li JJ, Zhang HQ, Li PJ, Xin ZL, Xi AQ, Zhuo-Ma, Ding YH, Yang ZP, Ma SQ
- 7043 Patients' awareness about their own breast cancer characteristics
Geng C, Lu GJ, Zhu J, Li YY
- 7053 Fracture risk assessment in children with benign bone lesions of long bones
Li HB, Ye WS, Shu Q

SYSTEMATIC REVIEWS

- 7062** Mothers' experiences of neonatal intensive care: A systematic review and implications for clinical practice
Wang LL, Ma JJ, Meng HH, Zhou J

META-ANALYSIS

- 7073** *Helicobacter pylori* infection and peptic ulcer disease in cirrhotic patients: An updated meta-analysis
Wei L, Ding HG

CASE REPORT

- 7085** Tuberous sclerosis complex-lymphangiomyomatosis involving several visceral organs: A case report
Chen HB, Xu XH, Yu CG, Wan MT, Feng CL, Zhao ZY, Mei DE, Chen JL
- 7092** Long-term survivor of metastatic squamous-cell head and neck carcinoma with occult primary after cetuximab-based chemotherapy: A case report
Große-Thie C, Maletzki C, Junghanss C, Schmidt K
- 7099** Genetic mutations associated with sensitivity to neoadjuvant chemotherapy in metastatic colon cancer: A case report and review of literature
Zhao L, Wang Q, Zhao SD, Zhou J, Jiang KW, Ye YJ, Wang S, Shen ZL
- 7110** Coexistence of cervical extramedullary plasmacytoma and squamous cell carcinoma: A case report
Zhang QY, Li TC, Lin J, He LL, Liu XY
- 7117** Reconstruction of the chest wall after resection of malignant peripheral nerve sheath tumor: A case report
Guo X, Wu WM, Wang L, Yang Y
- 7123** A rare occurrence of a hereditary Birt-Hogg-Dubé syndrome: A case report
Lu YR, Yuan Q, Liu J, Han X, Liu M, Liu QQ, Wang YG
- 7133** Late-onset Leigh syndrome without delayed development in China: A case report
Liang JM, Xin CJ, Wang GL, Wu XM
- 7139** New mechanism of partial duplication and deletion of chromosome 8: A case report
Jiang Y, Tang S, He F, Yuan JX, Zhang Z
- 7146** S-1 plus temozolomide as second-line treatment for neuroendocrine carcinoma of the breast: A case report
Wang X, Shi YF, Duan JH, Wang C, Tan HY
- 7154** Minimally invasive treatment of hepatic hemangioma by transcatheter arterial embolization combined with microwave ablation: A case report
Wang LZ, Wang KP, Mo JG, Wang GY, Jin C, Jiang H, Feng YF
- 7163** Progressive disfiguring facial masses with pupillary axis obstruction from Morbihan syndrome: A case report
Zhang L, Yan S, Pan L, Wu SF

- 7169** Idiopathic basal ganglia calcification associated with new *MYORG* mutation site: A case report
Fei BN, Su HZ, Yao XP, Ding J, Wang X
- 7175** Geleophysic dysplasia caused by a mutation in *FBNI*: A case report
Tao Y, Wei Q, Chen X, Nong GM
- 7181** Combined laparoscopic-endoscopic approach for gastric glomus tumor: A case report
Wang WH, Shen TT, Gao ZX, Zhang X, Zhai ZH, Li YL
- 7189** Aspirin-induced long-term tumor remission in hepatocellular carcinoma with adenomatous polyposis coli stop-gain mutation: A case report
Lin Q, Bai MJ, Wang HF, Wu XY, Huang MS, Li X
- 7196** Prenatal diagnosis of isolated lateral facial cleft by ultrasonography and three-dimensional printing: A case report
Song WL, Ma HO, Nan Y, Li YJ, Qi N, Zhang LY, Xu X, Wang YY
- 7205** Therapy-related myeloid leukemia during erlotinib treatment in a non-small cell lung cancer patient: A case report
Koo SM, Kim KU, Kim YK, Uh ST
- 7212** Pediatric schwannoma of the tongue: A case report and review of literature
Yun CB, Kim YM, Choi JS, Kim JW
- 7218** Status epilepticus as a complication after COVID-19 mRNA-1273 vaccine: A case report
Šin R, Štruncová D
- 7224** Successful outcome of retrograde pancreatojejunostomy for chronic pancreatitis and infected pancreatic cysts: A case report
Kimura K, Adachi E, Toyohara A, Omori S, Ezaki K, Ihara R, Higashi T, Ohgaki K, Ito S, Maehara SI, Nakamura T, Maehara Y
- 7231** Incidentally discovered asymptomatic splenic hamartoma misdiagnosed as an aneurysm: A case report
Cao XF, Yang LP, Fan SS, Wei Q, Lin XT, Zhang XY, Kong LQ
- 7237** Secondary peripheral T-cell lymphoma and acute myeloid leukemia after Burkitt lymphoma treatment: A case report
Huang L, Meng C, Liu D, Fu XJ
- 7245** Retroperitoneal bronchogenic cyst in suprarenal region treated by laparoscopic resection: A case report
Wu LD, Wen K, Cheng ZR, Alwalid O, Han P
- 7251** Coexistent vestibular schwannoma and meningioma in a patient without neurofibromatosis: A case report and review of literature
Zhao LY, Jiang YN, Wang YB, Bai Y, Sun Y, Li YQ
- 7261** Thoracoabdominal duplication with hematochezia as an onset symptom in a baby: A case report
Yang SB, Yang H, Zheng S, Chen G

- 7269 Dental management of a patient with Moebius syndrome: A case report
Chen B, Li LX, Zhou LL
- 7279 Epidural gas-containing pseudocyst leading to lumbar radiculopathy: A case report
Chen Y, Yu SD, Lu WZ, Ran JW, Yu KX
- 7285 Regression of intervertebral disc calcification combined with ossification of the posterior longitudinal ligament: A case report
Wang XD, Su XJ, Chen YK, Wang WG

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Idiopathic basal ganglia calcification associated with new MYORG mutation site: A case report

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Abstract

BACKGROUND

Idiopathic basal ganglia calcification (IBGC) is a neurodegenerative disease characterized by symmetrical calcification of basal ganglia and other brain region, also known as Fahr's disease. It can be sporadic or familial, and there is no definite etiology at present. With the development of neuroimaging, the number of reports of IBGC has increased in recent years. However, due to its hidden onset, diverse clinical manifestations, and low incidence, it is likely to be misdiagnosed or ignored by potential patients and their family.

CASE SUMMARY

We report a case of a 61-year-old man who presented with symptoms of dysphagia and alalia. His computed tomography scan of the brain revealed bilateral symmetric calcifications of basal ganglia, cerebellum, thalamus, and periventricular area. The genetic test showed a new mutation sites of MYORG, c.1438T>G mutation and c.1271_1272 TGGTGCGC insertion mutation. He was finally diagnosed with IBGC.

CONCLUSION

It is important to detect MYORG mutation when IBGC is suspected, especially in those without an obvious family history, for better understanding of the underlying mechanism and identifying potential treatments.

Key Words: Idiopathic basal ganglia calcification; Fahr's disease; Gene; Point mutation; Inheritance; Case report

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Core Tip: Idiopathic basal ganglia calcification (IBGC) is characterized by calcification of basal ganglia and other regions of the brain. IBGC is clinically heterogeneous and usually exhibits an autosomal dominant pattern of inheritance. We report a case of a 61-year-old man who presented with symptoms of dysphagia and alalia. His computed tomography scan of the brain revealed bilateral symmetric calcifications of basal ganglia, cerebellum, thalamus, and periventricular area. The genetic test showed a new mutation site of *MYORG*.

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INTRODUCTION

Idiopathic basal ganglia calcification (IBGC) is characterized by calcification of basal ganglia and other regions of the brain, commonly known as Fahr's disease[1]. IBGC is clinically heterogeneous and usually exhibits an autosomal dominant pattern of inheritance. Wang *et al*[2] discovered the first pathogenic gene of IBGC: *SLC20A2* in 2012. Three other IBGC pathogenic genes (*PDGFRB*[3], *PDGFB*[4], and *XPR1*[5]) were subsequently identified. In 2018, Yao *et al*[6] first reported that *MYORG* is a new pathogenic gene for IBGC with an autosomal-recessive trait. Here, we report a family with novel mutation sites of *MYORG*.

CASE PRESENTATION

Chief complaints

The patient is a 61-year-old man with the complaint of progressive severe dysphagia and alalia.

History of present illness

Patient's symptoms started 4 years ago and were getting worse. He denied any headache, seizures, and psychiatric symptoms.

History of past illness

Patient has hypertension for more than 10 years.

Personal and family history

Patient's parents passed away in their sixties with unknown reason. He had an old brother and three old sisters, three of them died of unknown reason. The surviving sister is clinically asymptomatic but has calcifications on computed tomography (CT) scan (Figure 1A), and total calcification[7] score was 10. His three daughters were also asymptomatic and with normal CT scan. The pedigree chart is shown in Figure 2.

Physical examination

The neurological examination was normal.

Laboratory examinations

Laboratory tests, including serum calcium, phosphorus, and parathyroid hormone levels, were within normal limits (calcium 2.22 mmol/L, phosphorus 0.84 mmol/L, parathyroid hormone 37.5 ng/L).

Imaging examinations

Patient's CT scan revealed multiple symmetric calcifications of bilateral basal ganglia,

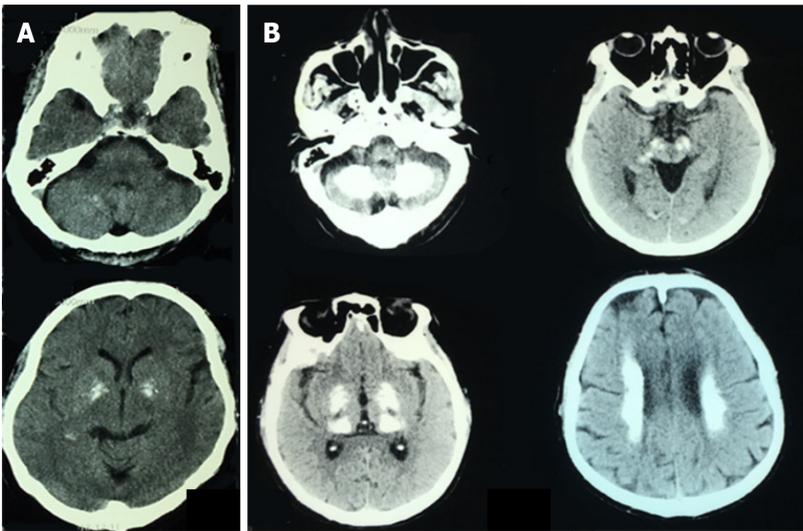


Figure 1 Computed tomography. A: Basal ganglia and cerebellum calcification of patient's sister; B: Patient's symmetric calcifications of bilateral basal ganglia, cerebellum, thalamus, and periventricular area.

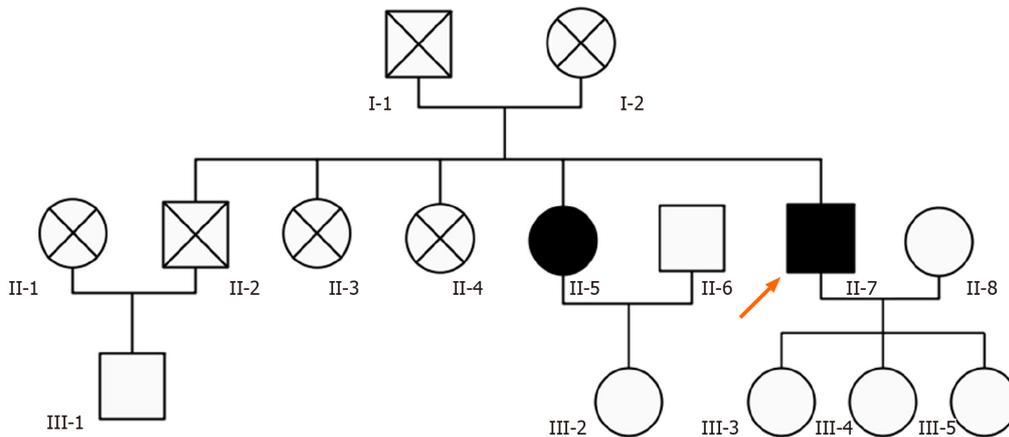


Figure 2 Pedigree. The patients with idiopathic basal ganglia calcification are represented in black, unaffected individuals in white. The index patient is indicated by an arrow.

cerebellum, thalamus, and periventricular area (Figure 1B). Total calcification score was 54.

Genetic tests

Genetic tests were done for the patient and his family. Excluding the dead and unreachable individuals, 7 people over two generations of the patient's family were investigated. The patient's genetic test showed c.1438T >G mutation and c.1271_1272 TGGTGCGC insertion mutation in exon 2 of MYORG gene. Besides the patient, his sister and two of his daughter have MYORG c.1438T>G mutation, while one daughter has MYORG c.1271_1272 TGGTGCGC insertion mutation (Table 1 and Figure 3).

FINAL DIAGNOSIS

According to these laboratory tests and neuroimaging, he was diagnosed with IBGC.

TREATMENT

Oxiracetam and nicergoline were given for neurotrophic treatments and improving cerebrovascular circulation.

Table 1 Clinical characteristics of pedigree

	II-5	II-7	II-8	III-1	III-3	III-4	III-5
Age at evaluation	66	61	61	51	36	34	32
Education level	II	II	II	III	IV	IV	IV
MMSE (/30)	27	26	28	29	30	30	30
MoCA (/30)	26	19	27	28	30	29	29
Clinic symptom	-	+	-	-	-	-	-
Calcifications on CT	+	+	-	-	-	-	-
Total calcification score	54	10	0	0	0	0	0
<i>MYORG</i> mutation	+	+	-	-	+	+	+
c.1438T>G mutation	-	+	-	-	-	-	+
c.1271_1272 TGGTGCGC insertion mutation	+	+	-	-	+	+	-

CT: Computed tomography; MMSE: Mini-mental state examination; MoCA: Montreal Cognitive Assessment.

OUTCOME AND FOLLOW-UP

The symptoms were not approved with these agents.

DISCUSSION

IBGC is a hereditary disease of the nervous system. With the wide application of CT, the detection rate of IBGC becomes higher and higher. Most patients were healthy in the adolescent period. Even if characteristic calcification lesions had appeared in the head CT examination, there might not be obvious abnormal symptoms of the nervous system. Neuropsychiatric and motor disorders would gradually present in the middle-aged and elderly patients with progressive parkinsonism and spasticity.

In this pedigree, the patient had compound heterozygous mutations, one point mutation and one insertion mutation. His sister and daughters only had one mutation site, and they did not present with any symptoms. Some research found that homozygotes have more serious brain calcification, whereas with the heterozygous mutation, only punctated calcification was found in the globus pallidus. The clinical symptoms in homozygotes are various neurological symptoms, while heterozygotes are usually clinically asymptomatic[8]. Although the patient's sister and daughters are asymptomatic, they had the *MYORG* mutation. We suggested to them annual cranial CT scans, because with age the disease may progress and clinical symptoms may appear.

Previous studies reported that IBGC is inherited as an autosomal dominant disease [9,10] and identified *SLC20A2* coding for the phosphate transporter PiT2, *PDGFRB*³ coding for the platelet derived growth factor receptor, as a cause of IBGC. However, *SLC20A2*, *PDGFRB*, *PDGFB*, and *XPR1* mutations were only detected in 16.8% of familial brain calcification. The underlying reason of IBGC remains unclear, especially in those without a prominent family history. In 2018, Yao *et al*[6] identified *MYORG* as the new recessive causal gene for IBGC, and reported 10 different mutation sites. Our patient and his family have *MYORG* novel mutation sites, c.1438T>G mutation and c.1271_1272 TGGTGCGC insertion mutation, which have not been reported previously.

MYORG, also known as KIAA1161 or NET37, is located on chromosome 9p13.3. It encodes a protein containing 714 amino acids, which is a member of the glycosylase 31 family. *MYORG* gene was specifically expressed in S100b positive astrocytes. Bilateral symmetrical nodules of cerebral calcification were observed in *MYORG* knockout mice. The main components of calcium nodules were calcium and phosphorus, which were highly similar to the elements of brain calcification in clinical patients, which proved that *MYORG* autosomal recessive function deletion mutation was an important pathogenic gene of IBGC. The mutation of this gene causes dysfunction of astrocytes, which may affect the combination of astrocytes and pericytes, interfere with the

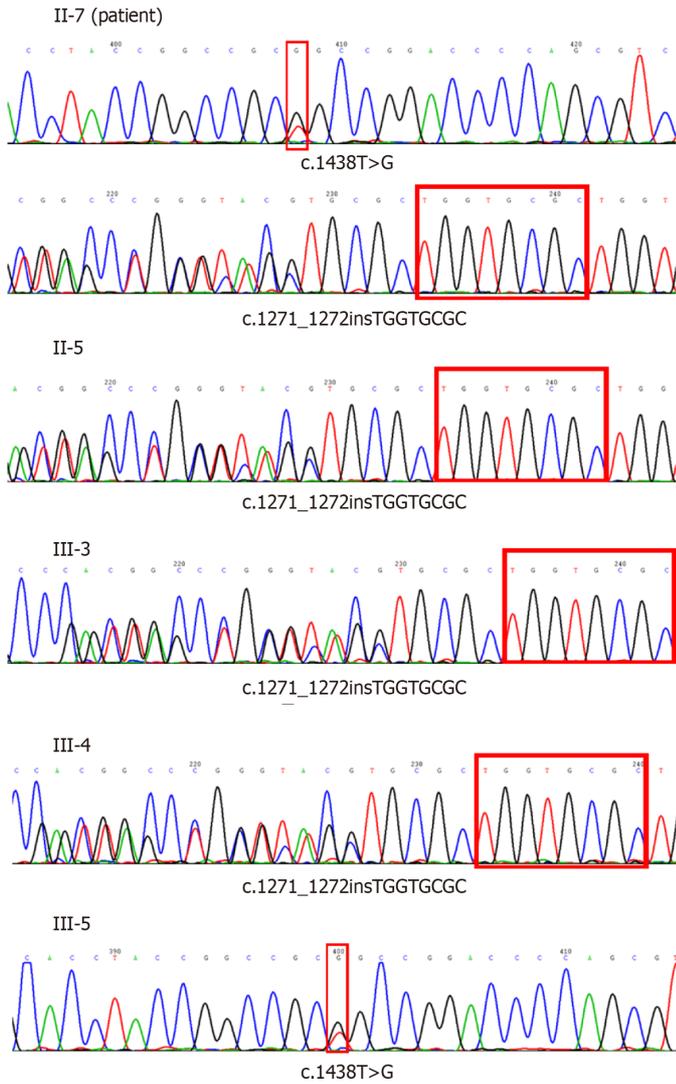


Figure 3 Gene sequencing. II-7 (patient) has c.1438T>G mutation and c.1271_1272 TGGTGCGC insertion mutation. II-5, III-3 and III-4 has c.1271_1272 TGGTGCGC insertion mutation. III-5 has c.1438T>G mutation.

normal function of neurovascular unit, and finally may lead to the calcification of the brain. According to the clinical manifestations, Chen *et al*[11] found that 76.5% patients with MYORG mutation were symptomatic, and the most recurrent symptom was movement disorder. Our patient’s manifestation was progressive dysphagia and alalia.

CONCLUSION

Since many patients will exhibit a progressive neurological disorder with no known treatment, it is important to detect MYORG mutation, especially in those without obvious family history, for better understanding the underlying mechanism and identifying potential treatments.

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