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Contents Thrice Monthly Volume 10 Number 23 August 16, 2022

OPINION REVIEW

8057 Invasive intervention timing for infected necrotizing pancreatitis: Late invasive intervention is not late for collection

Xiao NJ, Cui TT, Liu F, Li W

8063 Clinical utility of left atrial strain in predicting atrial fibrillation recurrence after catheter ablation: An up-to-date review

Yu ZX, Yang W, Yin WS, Peng KX, Pan YL, Chen WW, Du BB, He YQ, Yang P

MINIREVIEWS

8076 Gut microbiota and COVID-19: An intriguing pediatric perspective

Valentino MS, Esposito C, Colosimo S, Caprio AM, Puzone S, Guarino S, Marzuillo P, Miraglia del Giudice E, Di Sessa A

8088 Beta receptor blocker therapy for the elderly in the COVID-19 era

Santillo E, Migale M

ORIGINAL ARTICLE

Retrospective Cohort Study

8097 Nonselective beta-blocker use is associated with increased hepatic encephalopathy-related readmissions in cirrhosis

Fallahzadeh MA, Asrani SK, Tapper EB, Saracino G, Rahimi RS

Retrospective Study

8107 Different squatting positions after total knee arthroplasty: A retrospective study

Li TJ, Sun JY, Du YQ, Shen JM, Zhang BH, Zhou YG

8115 Outcomes of seromuscular bladder augmentation compared with standard bladder augmentation in the treatment of children with neurogenic bladder

Sun XG, Li YX, Ji LF, Xu JL, Chen WX, Wang RY

8124 Distinctive clinical features of spontaneous pneumoperitoneum in neonates: A retrospective analysis

Kim SH, Cho YH, Kim HY

8133 Cognitive training for elderly patients with early Alzheimer's disease in the Qinghai-Tibet Plateau: A pilot study

Wang XH, Luo MQ

8141 Diagnostic value of elevated serum carbohydrate antigen 125 level in sarcoidosis

Zhang Q, Jing XY, Yang XY, Xu ZJ

World Journal of Clinical Cases

Thrice Monthly Volume 10 Number 23 August 16, 2022

Contents

8152 Evaluation of progressive early rehabilitation training mode in intensive care unit patients with mechanical ventilation

Qie XJ, Liu ZH, Guo LM

8161 Comparison of demographic features and laboratory parameters between COVID-19 deceased patients and surviving severe and critically ill cases

Wang L, Gao Y, Zhang ZJ, Pan CK, Wang Y, Zhu YC, Qi YP, Xie FJ, Du X, Li NN, Chen PF, Yue CS, Wu JH, Wang XT, Tang YJ, Lai QQ, Kang K

Clinical Trials Study

8170 Role of H_2 receptor blocker famotidine over the clinical recovery of COVID-19 patients: A randomized controlled trial

Mohiuddin Chowdhury ATM, Kamal A, Abbas MKU, Karim MR, Ali MA, Talukder S, Hamidullah Mehedi H, Hassan H, Shahin AH, Li Y, He S

Observational Study

8186 Short-term prognostic factors for hepatitis B virus-related acute-on-chronic liver failure

Ye QX, Huang JF, Xu ZJ, Yan YY, Yan Y, Liu LG

8196 Three-dimensional psychological guidance combined with evidence-based health intervention in patients with liver abscess treated with ultrasound

Shan YN, Yu Y, Zhao YH, Tang LL, Chen XM

8205 Role of serum β2-microglobulin, glycosylated hemoglobin, and vascular endothelial growth factor levels in diabetic nephropathy

Yang B, Zhao XH, Ma GB

SYSTEMATIC REVIEWS

8212 Gallbladder neuroendocrine carcinoma diagnosis, treatment and prognosis based on the SEER database: A literature review

Cai XC, Wu SD

CASE REPORT

8224 Sepsis complicated with secondary hemophagocytic syndrome induced by giant gouty tophi rupture: A case report

Lai B, Pang ZH

8232 Spontaneous remission of autoimmune pancreatitis: Four case reports

Zhang BB, Huo JW, Yang ZH, Wang ZC, Jin EH

8242 Epstein–Barr-virus-associated hepatitis with aplastic anemia: A case report

Zhang WJ, Wu LQ, Wang J, Lin SY, Wang B

8249 Aspiration as the first-choice procedure for airway management in an infant with large epiglottic cysts: A case report

Zheng JQ, Du L, Zhang WY

World Journal of Clinical Cases

Thrice Monthly Volume 10 Number 23 August 16, 2022

Contents

8255 Sequential multidisciplinary minimally invasive therapeutic strategy for heart failure caused by four diseases: A case report

Zhao CZ, Yan Y, Cui Y, Zhu N, Ding XY

8262 Primary ascending colon cancer accompanying skip metastases in left shoulder skin and left neck lymph node: A case report

Zhou JC, Wang JJ, Liu T, Tong Q, Fang YJ, Wu ZQ, Hong Q

8271 Clinical and genetic study of ataxia with vitamin E deficiency: A case report

Zhang LW, Liu B, Peng DT

8277 Complete resection of large-cell neuroendocrine and hepatocellular carcinoma of the liver: A case report

Noh BG, Seo HI, Park YM, Kim S, Hong SB, Lee SJ

8284 Immunotherapy combined with antiangiogenic agents in patients with advanced malignant pleural mesothelioma: A case report

Xuan TT, Li GY, Meng SB, Wang ZM, Qu LL

8291 Bladder malacoplakia: A case report

Wang HK, Hang G, Wang YY, Wen Q, Chen B

8298 Delayed inflammatory response evoked in nasal alloplastic implants after COVID-19 vaccination: A case report

Seo MG, Choi EK, Chung KJ

8304 Phosphoglyceride crystal deposition disease requiring differential diagnosis from malignant tumors and confirmed by Raman spectroscopy: A case report

Ohkura Y, Uruga H, Shiiba M, Ito S, Shimoyama H, Ishihara M, Ueno M, Udagawa H

8312 Vulvovaginal myeloid sarcoma with massive pelvic floor infiltration: A case report and review of literature

Wang JX, Zhang H, Ning G, Bao L

8323 Femoral neck stress fracture and medial tibial stress syndrome following high intensity interval training: A case report and review of literature

Tan DS, Cheung FM, Ng D, Cheung TLA

8330 Periosteal chondroma of the rib: A case report

Gao Y, Wang JG, Liu H, Gao CP

8336 Papillary thyroid carcinoma occurring with undifferentiated pleomorphic sarcoma: A case report

Lee YL, Cheng YQ, Zhu CF, Huo HZ

8344 Laparoscopic treatment of bilateral duplex kidney and ectopic ureter: A case report

Wang SB, Wan L, Wang Y, Yi ZJ, Xiao C, Cao JZ, Liu XY, Tang RP, Luo Y

8352 Incontinentia pigmenti with intracranial arachnoid cyst: A case report

Li WC, Li ML, Ding JW, Wang L, Wang SR, Wang YY, Xiao LF, Sun T

8360 Relapsing polychondritis causing breathlessness: Two case reports

Zhai SY, Zhang YH, Guo RY, Hao JW, Wen SX

8367 Endodontic management of a fused left maxillary second molar and two paramolars using cone beam computed tomography: A case report

Mei XH, Liu J, Wang W, Zhang QX, Hong T, Bai SZ, Cheng XG, Tian Y, Jiang WK

8375 Infant biliary cirrhosis secondary to a biliary inflammatory myofibroblastic tumor: A case report and review of literature

Huang Y, Shu SN, Zhou H, Liu LL, Fang F

8384 Metastatic low-grade endometrial stromal sarcoma with variable morphologies in the ovaries and mesentery: A case report

Yu HY, Jin YL

8392 Bronchogenic cysts with infection in the chest wall skin of a 64-year-old asymptomatic patient: A case report

Ma B, Fu KW, Xie XD, Cheng Y, Wang SQ

8400 Incidental accumulation of Technetium-99m pertechnetate in subacute cerebral infarction: A case report

Han YH, Jeong HJ, Kang HG, Lim ST

8406 Metal stent combined with ileus drainage tube for the treatment of delayed rectal perforation: A case report

Cheng SL, Xie L, Wu HW, Zhang XF, Lou LL, Shen HZ

8417 Using ketamine in a patient with a near-occlusion tracheal tumor undergoing tracheal resection and reconstruction: A case report

Xu XH, Gao H, Chen XM, Ma HB, Huang YG

LETTER TO THE EDITOR

8422 Reflections on the prevalence of human leukocyte antigen-B27 and human leukocyte antigen-B51 co-occurrence in patients with spondylarthritis

Gonçalves Júnior J, Sampaio-Barros PD, Shinjo SK

8425 Comment on "Disease exacerbation is common in inflammatory bowel disease patients treated with immune checkpoint inhibitors for malignancy"

Argyriou K, Kotsakis A

8428 Intranasal sufentanil combined with intranasal dexmedetomidine: A promising method for non-anesthesiologist sedation during endoscopic ultrasonography

Wang Y, Ge ZJ, Han C

World Journal of Clinical Cases

Thrice Monthly Volume 10 Number 23 August 16, 2022

Contents

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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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The *WJCC* is now abstracted and indexed in Science Citation Index Expanded (SCIE, also known as SciSearch®), Journal Citation Reports/Science Edition, Current Contents®/Clinical Medicine, PubMed, PubMed Central, Scopus, Reference Citation Analysis, China National Knowledge Infrastructure, China Science and Technology Journal Database, and Superstar Journals Database. The 2022 Edition of Journal Citation Reports® cites the 2021 impact factor (IF) for *WJCC* as 1.534; IF without journal self cites: 1.491; 5-year IF: 1.599; Journal Citation Indicator: 0.28; Ranking: 135 among 172 journals in medicine, general and internal; and Quartile category: Q4. The *WJCC*'s CiteScore for 2021 is 1.2 and Scopus CiteScore rank 2021: General Medicine is 443/826.

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CASE REPORT

Clinical and genetic study of ataxia with vitamin E deficiency: A case report

Lin-Wei Zhang, Bing Liu, Dan-Tao Peng

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Abstract

BACKGROUND

Ataxia with vitamin E deficiency (AVED) is a type of autosomal recessive cerebellar ataxia. Clinical manifestations include progressive cerebellar ataxia and movement disorders. *TTPA* gene mutations cause the disease.

CASE SUMMARY

We report the case of a 32-year-old woman who presented with progressive cerebellar ataxia, dysarthria, dystonic tremors and a remarkably decreased serum vitamin E concentration. Brain magnetic resonance images showed that her brainstem and cerebellum were within normal limits. Acquired causes of ataxia were excluded. Whole exome sequencing subsequently identified a novel homozygous variant (c.473T>C, p.F158S) of the *TPPA* gene. Bioinformatic analysis predicted that F185S is harmful to protein function. After supplementing the patient with vitamin E 400 mg three times per day for 2 years, her symptoms remained stable.

CONCLUSION

We identified an AVED patient caused by novel mutation in *TTPA* gene. Our findings widen the known *TTPA* gene mutation spectrum.

Key Words: Ataxia with vitamin E deficiency; *TTPA* gene; Tremor; Case report

Core Tip: Ataxia with vitamin E deficiency can present as progressive chronic cerebellar ataxia and involuntary movement disorder. Vitamin E supplementation should be initiated as early as possible to stop disease progression.

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INTRODUCTION

Ataxia with vitamin E deficiency (AVED) is a type of autosomal recessive cerebellar ataxia (ARCA). The most prominent symptoms include cerebellar ataxia, areflexia, peripheral neuropathy and movement disorder[1,2]. AVED patients may also exhibit retinitis pigmentosa, cardiomyopathy and scoliosis[3,4]. AVED is caused by mutations in the *TTPA* gene, which encodes the α-tocopherol transfer protein, which in turn binds α-tocopherol and transports vitamin E from hepatocytes to circulating lipoproteins[5]. Vitamin E supplementation may prevent the worsening of the condition of patients with AVED.

To date, the known incidence rate of AVED in China has been low[6]. Here, we report the clinical, biochemical and genetic investigation of a Chinese patient with AVED due to homozygous mutations in the *TTPA* gene. This patient exhibited progressive cerebellar ataxia, dysarthria and head titubation with markedly low levels of serum vitamin E. After treatment with vitamin E 400 mg three times per day for 2 years, the patient's neurological symptoms were stabilized.

CASE PRESENTATION

Chief complaints

A 32-year-old woman was admitted to the hospital because of progressive cerebellar ataxia and involuntary head tremors.

History of present illness

When the patient was 14-years-old, she began exhibiting an unsteady gait which worsened to become wide-based and staggering. She also developed slurring of speech and clumsiness of the hands. At 18 years of age, she began having involuntary dystonic head tremors as well.

History of past illness

She had no specific intestinal lesions indicative of malabsorption. Past medical history was unremarkable.

Personal and family history

Her parents are cousins. There is no family history of similar neurological disorders.

Physical examination

Neurological examination revealed normal cognitive state, dysarthria, overt head tremor, bilateral dysmetria on nose-finger and heel-shin tests and a wide-based ataxic gait with inability to walk in tandem. Kayser-Fleischer Rings were absent; vision and hearing ability were normal. Motor and sensory examinations yielded normal results apart from areflexia. Romberg's sign and bilateral Babinski sign were positive; Pes cavus deformity noted. Obvious overt head dystonic tremor, bilateral dysmetria on finger-to- nose and heel-to-shin tests and wide-based gait with inability to walk in tandem. Scale for the assessment and rating of ataxia (SARA) score was 11 [gait 2, stance 2, sitting 0, speech 1, finger chase 1.0 (left 1, right 1), nose-finger test 2.0 (left 2, right 2), fast alternating hand movements 1.0 (left 1, right 1), and heel-shin slide 2.0 (left 2, right 2)].

Laboratory examinations

Routine blood tests, including liver function, autoimmune antibodies, thyroid function, blood smear for acanthocytosis and plasma levels of vitamins (B1, B2, B6, B9, B12, A, D, E), copper and ceruloplasmin were normal. Cerebrospinal fluid was normal including inflammatory, immunological and infectious indices. Electromyography, nerve conduction velocity and brainstem auditory evoked potential were all normal. Initial DNA analyses using capillary electrophoresis of polymerase chain reaction products excluded Friedrich's ataxia, spinocerebellar ataxia (1, 2, 3, 6, 7, 8, 10, 12, 17) and dentatorubral-pallid-

oluysian atrophy. The plasma level of total vitamin E detected *via* high performance liquid chromatography was 0.59 μg/mL (normal: 10.8 ± 3.3 mg/L[7]).

Imaging examinations

Brain magnetic resonance imaging (MRI) showed no obvious atrophy of brainstem and cerebellum (Figure 1).

Further diagnostic work-up

Genomic DNA was extracted from peripheral leukocytes of the patient and all available family members who gave written informed consent, according to the standard protocol approved by the China-Japan Friendship Hospital. DNA of the proband was subjected to whole exome sequencing (WES) using the Ion Torrent AmpliSeq Exome RDY kit (BGI Tech, Hong Kong). Variant call files were analyzed with Ingenuity Variant Analysis (Qiagen, Redwood City, CA, United States) using an autosomal recessive model. Clean reads were aligned on the human assembly GRCh37 by Burrow-Wheeler aligner. Small insertions/deletions and single nucleotide variants were called by genomic analysis toolkit and annotated by ANNOVAR. Several filtration steps to obtain putative pathogenic variants were processed. The functional effects of protein variants were predicted by Sorting Intolerant from Tolerant (SIFT), PolyPhen2 and MutationTaster. Disease association databases (*e.g.*, HGMD, OMIM and ClinVar) and genetic variation databases (*e.g.*, 1000 Genomes Project, ESP6500 and ExAC) were used in the filtering process as well. Potential pathogenic variants were validated by conventional Sanger sequencing and her family members were included for segregation analysis. We used the transcript sequence (OMIM*600415, NM_000370.3) of the *TTPA* gene and discovered homozygous variants(c.473C>T, p.Phe185Ser) in the proband. Sanger sequencing confirmed this result and revealed that her parents and the younger sister are heterozygous carriers for 473C>T (Figure 2). This mutation was predicted to be harmful (SIFT: tolerated, Polyphen2: possibly damaging, MutationTaster: disease causing). The 158-phenylalanine residue affected by the mutation were highly conserved in evolution (Figure 3).

FINAL DIAGNOSIS

The final diagnosis of the presented case was AVED due to *TTPA* homozygous missense mutation (c.473C>T, p.Phe185Ser) (GRCh37/hg19).

TREATMENT

After we detected decreased plasma levels of total vitamin E, supplementation with vitamin E 400 mg three times per day was immediately administered. The patient also kept physical exercises for rehabilitation therapy.

OUTCOME AND FOLLOW-UP

After 2 years of vitamin E supplementation therapy, the symptoms of this patient showed neither improvement nor deterioration. On her last follow-up visit, her SARA score was 11 [gait 2, stance 2, sitting 0, speech 1, finger chase 1.5 (left 2, right 1), nose-finger test 1.5 (left 2, right 1), fast alternating hand movements 1.0 (left 1, right 1) and heel-shin slide 2.0 (left 2, right 2)].

DISCUSSION

By WES we identified homozygous mutations of *TTPA* gene (c.473C>T, p.Phe185Ser) in a Chinese family with ARCA and we have excluded common causes of ataxia. AVED was first reported by Burck *et al*[8]. It usually manifests as a mild disease course and is rarely reported in China. The presentation of AVED is highly heterogeneous with onset usually in childhood[9] but some cases have manifested in infancy and in adulthood[1]. Clinical phenotypes include progressive gait ataxia, movement disorders, areflexia, dysarthria, epilepsy, pyramidal signs, impaired proprioception and vibration sense and sensory neuropathy[9-11]. Apart from cerebellar ataxia, our patient exhibited obvious head tremor. Head titubation, seen in 37% to 73% of patients, and cervical dystonia are distinguishing motor features of AVED[1,3,12]. Non-neurological symptoms, such as retinitis pigmentosa, macular degeneration and cardiomyopathy[13,14] were not found in our patient. Laboratory examination often reveals markedly low serum vitamin E concentration in AVED[1]. The patient's MRI showed no obvious cerebellar

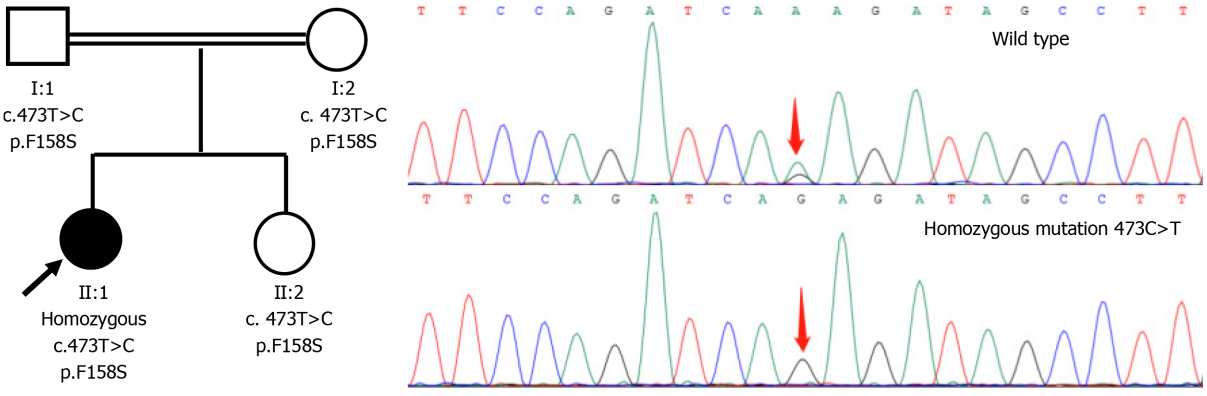

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Figure 1 Pedigree and sanger sequencing validation of homozygous mutation c.473T>C, p.F158S of *TTPA* gene in this pedigree. Clear square: Male; Clear circle: Female; Black circle: Index patient.

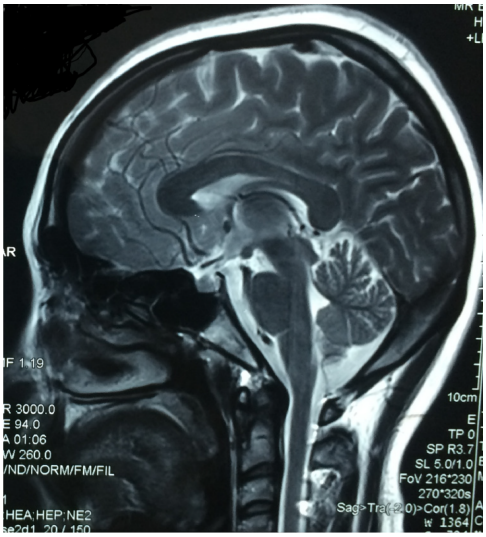

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Figure 2 Brain magnetic resonance images show relatively normal brainstem and cerebellum of the index patient (II:1).

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Figure 3 Evaluation of the mutation p.Arg301Gln is highly conserved among species.

atrophy in our patient and absence of cerebellar atrophy is common in AVED patients[15].

To date, various mutations of the *TTPA* gene have been reported[16]. The genotype-phenotype correlations were not found to be strong in AVED and c.744delA was the most frequent mutation, often originating in the Mediterranean region[9]. Moreover, 744delA could increase the risk of early age onset, severe disease course and cardiomyopathy[17]. c.473C>T(p.F185S), which was detected in our patient, has not been reported previously. Her symptoms began in adolescence and she is still able to walk 18 years after the onset of disease. F185S is located in CRAL_TRIO domain and its predicted function is to

combine with α-tocopherol[18]. Bioinformatic analysis predicted this novel mutation would lead to the damage of protein function.

AVED is a treatable form of hereditary ataxia and early and sustained vitamin E supplementation could result in a remarkable clinical response or stabilization in AVED patients[1,19,20]. In patients with ataxia, a prompt investigation for vitamin E deficiency is recommended[19,20]. AVED patients require lifelong vitamin E supplementation at 300-2400 mg/d to maintain adequate plasma levels of vitamin E [1,8,14]. Our patient received 1200 mg/d of vitamin E for 2 years and her symptoms showed no deterioration.

CONCLUSION

We identified a Chinese AVED female patient caused by a novel mutation in the *TTPA* gene. This finding widens the known *TTPA* gene mutation spectrum.

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FOOTNOTES

Author contributions: Zhang LW contributed to acquisition of data, carried out the molecular genetic studies, analyzed the molecular and clinical data, wrote the main manuscript text and prepared figures; Liu B collected data during the study; Peng DT critically revised the manuscript for important intellectual content and supervised the study; All authors read and approved the manuscript.

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