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Transient diabetes mellitus with ABCC8 variant successfully treated with sulfonylurea: Two case reports and review of literature

Ling-Hua Shen, Yan Cui, Dong-Xia Fu, Wei Yang, Sheng-Nan Wu, Hui-Zhen Wang, Hai-Hua Yang, Yong-Xing Chen, Hai-Yan Wei

Abstract

BACKGROUND
Transient neonatal diabetes mellitus (TNDM) is a rare form of diabetes mellitus that usually presents within the first 6 mo of life. Patients often enter remission within several months, although relapse can occur later in life. Mutations in the ABCC8 gene, which encodes the sulfonylurea receptor 1 of the ATP-sensitive potassium channel in pancreatic beta cells, are associated with TNDM and permanent neonatal diabetes. This study describes a novel de novo c.3880C>T heterozygous ABCC8 variant that causes TNDM and can be treated with sulfonylurea therapy.

CASE SUMMARY
We retrospectively analyzed 2 Chinese patients with TNDM who were diagnosed, treated, or referred for follow-up between September 2017 and September 2023. The patients were tested for mutations using targeted next-generation sequencing. Patients with neonatal diabetes mellitus caused by a c.3880C>T heterozygous missense variant in the ABCC8 gene have not been reported before. Both children had an onset of post-infectious diabetic ketoacidosis, which is worth noting. At a follow-up visit after discontinuing insulin injection, oral glyburide was found to be effective with no adverse reactions.

CONCLUSION
Early genetic testing of neonatal diabetes mellitus aids in accurate diagnosis and treatment and helps avoid daily insulin injections that may cause pain.
INTRODUCTION

Neonatal diabetes mellitus (NDM) is characterized by the onset of diabetes within the first 6 mo of life, but some cases are diagnosed between 6 mo and 12 mo of age. NDM is a type of monogenic diabetes that is very rare and difficult to treat, as reported by Kitsell in 1851[7]. The incidence of NDM is estimated to be approximately 1/89000-1:50000 live births[8], but it is not known in China. There are two clinical NDM subtypes depending on the permanence of hyperglycemia, transient (TNDM) and permanent (PNDM)[9]. The latter is a lifelong disease without remission; but TNDM, which accounts for 50%-60% of the NDM cases, usually enters remission after 6-12 mo. However, such patients may relapse in adolescence or early adulthood[10]. There are over 20 known genetic causes of NDM[11]. Approximately two-thirds of TNDM cases are related to abnormalities in an imprinted region on chromosome 6q24. Activating mutations in either of the genes encoding the two subunits of the ATP-sensitive potassium (KATP) channel of the pancreatic β-cell membrane [potassium inwardly rectifying channel subfamily j member 11 (KCNJ11) or ABCC8] are responsible in most of the remaining cases (KATP-NDM)[12,13]. Patients with KATP-NDM respond to sulfonylurea (SU) therapy, and approximately 90%-95% may be successfully transitioned to SU therapy, with complete discontinuation of insulin and a significant decrease in glycated hemoglobin levels[14,15]. In China, reports of the ABCC8 gene variant causing TNDM are rare. Here we describe a clinical phenotype of TNDM caused by a novel de novo ABCC8 gene c.3880C>T heterozygous variant and report effective SU treatment in 2 Chinese infants.

CASE PRESENTATION

Chief complaints

Case 1: A male infant 2 mo and 14 d of age was admitted to our hospital following a fever of 4 d, dyspnea of 3 d, and high blood glucose of 1 d duration.

Case 2: A male infant 7 mo and 20 d of age was admitted to our hospital following polydipsia and polyuria of 13 d, intermittent fever 6 d, and poor spirits of 5 d duration.

History of present illness

Case 1: Four days before admission, the parents complained of fever and increased water intake; however, the specific urine volume was not known. They reported that no chills, convulsions, nasal congestion, runny nose, cough, vomiting, abdominal distension, diarrhea, abnormal crying, or other symptoms had occurred. The patient had been given oral medication for a respiratory infection at a local clinic for 1 d. However, recurrent fever persisted. 3 d before admission he had dyspnea and was treated at a local hospital. But the response to atomization and oral medication was poor. One day earlier, he had an occasional cough and sputum in his throat accompanied by dyspnea, poor milk intake, poor spirits, and drowsiness. He was transferred to a superior local hospital for treatment. His blood glucose was high, and blood gas analysis revealed a pH of 7.06, a bicarbonate( HCO3-) level of 3.4 mmol/L, and a base excess(BE) of −24.8 mmol/L. The patient was treated with antibiotics, fluid replacement, and insulin therapy for 1 d. His breathing improved, and his body temperature was normal, but his blood glucose level was not well controlled. Therefore, he was transferred to our hospital for further treatment.
**Case 2:** Thirteen days before admission, the patient developed polydipsia and polyuria without evident cause, and the water intake and urine volumes were not known. Increased nocturia, evident overeating, or weight loss were not noted, but 6 d before admission, he had a fever with a peak temperature of 38 °C and occasional coughing. Oral ibuprofen effectively normalized the body temperature but was accompanied by frequent episodes of crying and agitation. He was taken to a local hospital and was treated with oral drugs for bronchitis. Furthermore, 5 d before admission, he manifested a poor spirit, mouth breathing, and tachypnea, with no nasal congestion, runny nose, cough, expectoration, rash, or convulsions. On a hospital revisit, he was considered to have severe pneumonia or respiratory failure. After receiving intravenous diazepam and treatment with cardiotonic drugs, his heart rate increased to 180-190 bpm, and he was transferred from the local facility to the intensive care unit of a tertiary hospital. His blood glucose was high, and blood gas analysis revealed a pH of 7.12, a HCO3 of 2.1 mmol/L, and a BE of −27 mmol/L. In addition, ventilator-assisted breathing, oxygen inhalation, sedation, anti-infectives, rehydration therapy, and other symptomatic treatments were administered. Insulin was given for 3 h but blood glucose levels remained at > 30 mmol/L. After the patient’s condition stabilized, an insulin pump was implanted for continuous subcutaneous insulin infusion. During hospitalization, the patient had an intermittent fever with a peak of 38.5 °C, accompanied by a cough and expectoration. 2 d before admission, the treatment was switched to regular subcutaneous insulin injection of 1.2 IU every 6 h. The blood glucose level fluctuated between 4-19 mmol/L, and the patient’s mental state and respiration were significantly improved. Subsequently, the patient was transferred to our hospital for further treatment.

**History of past illness**

**Cases 1 and 2:** The patients had no specific medical histories.

**Personal and family history**

**Case 1:** The patient was born at term by cesarean section, with a birth weight of 3.85 kg. The parents denied a history of asphyxia rescue. He was breastfed after birth, and his parents were not consanguineous. No family history of diabetes was reported.

**Case 2:** The patient was the second live birth of the mother (gravida 2, para 2), was delivered uneventfully at 38 wk, with a birth weight of 3 kg. The parents denied a history of asphyxia rescue. He was breastfed after birth and was not given complementary food. His parents were not consanguineous. No family history of diabetes was reported.

**Physical examination**

**Case 1:** Physical examination at admission showed a body temperature of 36.4 °C, pulse rate of 168 bpm, a respiratory rate of 42 breaths/min, blood pressure of 75/40 mmHg, height of 52 cm, and a weight of 7 kg. There was a poor spirit, no jaundice, no bleeding spots, normal skin elasticity, soft neck, normal fontanelle, equal pupils, sensitivity to light reflex, ruddy lips, smooth oral mucosa, rapid and regular breathing with thick breath sounds in both lungs, strong heart sounds, normal abdomen, normal spine and limbs, normal muscle strength, but poor peripheral circulation.

**Case 2:** Physical examination at admission showed a body temperature of 36.5 °C, pulse rate of 108 bpm, respiratory rate of 25 breaths/min, blood pressure of 80/45 mmHg, height of 66.8 cm (3rd-10th percentile), and weight of 10 kg. Clear consciousness, moderate nutrition, anemic appearance, pale complexion, no jaundice, no bleeding spots, normal skin elasticity, equal and round pupils, sensitivity to light reflex, soft neck, smooth breathing, thick breath sounds, and phlegm rales in both lungs, strong heart sounds, normal abdomen, normal spine and limbs, normal muscle strength, normal reflexes, slightly pale nail beds.

**Laboratory examination**

**Case 1:** Laboratory examinations were performed at admission. The venous blood glucose level was 11.15 mmol/L after admission and multiple random peripheral blood glucose levels were > 11.1 mmol/L. Blood gas analysis revealed a pH of 7.356, a HCO3 of 19.9 mmol/L, and a BE of 4.5 mmol/L. Urine glucose was markedly elevated (++++) and there was a weak positive result for urine ketone bodies. Blood ammonia and lactic acid levels were 67.8 μmol/L and 3.3 mmol/L, respectively. Routine blood tests revealed a white blood cell (WBC) count of 10.14 × 10^9/L, red blood cell (RBC) count of 3.3 × 10^12/L, Hb level of 99 g/L, and a platelet (PLT) count of 101 × 10^9/L. Blood biochemistry results were total cholesterol 6.27 mmol/L (reference range 0.36-6.2 mmol/L), triglycerides 2.46 mmol/L (reference range 0.38-2.25 mmol/L), and normal liver function and renal function, myocardial enzyme levels, electrolyte levels, and thyroid function results were normal. The C-peptide level was 0.88 ng/mL (reference range, 1.1-4.4 ng/mL), and glycosylated Hb (HbA1c) was 9.52%. Insulinulin autoantibodies (glutamic acid decarboxylase, islet cell, and insulin antibodies) were negative.

**Case 2:** Laboratory examination was performed at admission. The venous blood glucose level was 13.26 mmol/L, and multiple random peripheral blood glucose samples were > 11.1 mmol/L. Blood gas analysis revealed a pH of 7.411, a HCO3 of 24.5 mmol/L, and a BE of 0.4 mmol/L. Urine glucose and urine ketone bodies were weakly positive, respectively. The blood ammonia level was 15.5 μmol/L and the lactic acid level was 2.4 mmol/L. Routine blood tests showed a WBC count of 11.57 × 10^9/L, RBC count of 4.14 × 10^12/L, Hb level of 66 g/L, and PLT count of 317 × 10^9/L. Blood biochemistry results were triglycerides 3.61 mmol/L (reference range, 0.38-2.25 mmol/L) and normal liver function, renal function, myocardial enzymes, electrolytes, and thyroid function. The C-peptide level was 1.44 ng/mL (reference range, 1.1-4.4 ng/mL), HbA1c was 12.63%; and five insulin antibodies (glutamic acid decarboxylase autoantibodies, islet cell antibodies, insulin autoantibody, zinc transporter-8 antibody, and tyrosine phosphatase...
Genetic testing and mutation analysis: The Ethics Committee of the Children’s Hospital Affiliated with Zhengzhou University approved this study (Approval No. 2023-K-123). Informed consent was obtained from the patients’ guardians. DNA samples were sent to the hospital’s Institute of Pediatrics for next-generation sequencing. The procedure was performed following the standard protocol. Sanger sequencing was used to verify the variant sites in the patients, and sequence analysis was performed on samples from the parents to determine the source of variation. A heterozygous missense variant in ABCC8 was found in both patients, resulting in a C>T variant at position 3880 and the substitution of leucine (L) by phenylalanine (F) at amino acid position 1294 of the encoded protein. No corresponding gene variants were detected in the parents of either infant (Figure 1).

The ABCC8 c.3880C>T variant has not been reported in PubMed or other databases such as the Human Gene Mutation Database, ClinVar, and the Database for Single Nucleotide Polymorphisms (dbSNP). Homologous sequence comparison using DNAMAN software (Lynnon Biosoft Bioinformatics Solutions, San Ramon, CA, United States) showed that amino acid L at position 1294 of the ABCC8 protein was highly conserved (Figure 2). Bioinformatics software, such as Mutation Taster (https://www.mutationtaster.org/), PolyPhen-2 (http://genetics.bwh.harvard.edu/pph2), and SIFT (http://sift.bii.a-star.edu.sg/), predicted that the c.3880C>T variant was pathogenic or likely pathogenic. This variant changed the nonpolar hydrophobic aliphatic L to a nonpolar hydrophobic aromatic F at position 1294, altering the side chain structure with insignificant changes in hydrogen bonding (Figure 3). The c.3880C>T variant is classified as pathogenic (PM1 + PM2 + PP3 + PP4) by the American College of Medical Genetics and Genomics (ACMG) Classification of Genetic Variations[16].
Shen LH et al. ABCC8 variant-related TNDM

Figure 3 Predicted three-dimensional structure of the protein at c.3880C>T variant points of the ABCC8 gene. A: Three-dimensional structure of the wild-type protein at the 1294 position is that of leucine; B: C.3880C>T variant protein becomes phenylalanine, altering the side chain structure, with no significant change in the hydrogen bonding.

FINAL DIAGNOSIS

The final diagnosis was TNDM.

TREATMENT

Both infants were treated with low-dose insulin after diabetic ketoacidosis. The initial dose was as follows: after ketoacidosis was corrected, neutral protamine Hagedorn insulin was administered three times, and glyburide was used as an experimental treatment when the blood glucose level was relatively stable. The initial dose of glyburide was 0.1-0.2 mg/kg/day. The glyburide dose was gradually adjusted by the results of blood glucose monitoring and insulin was gradually discontinued.

OUTCOME AND FOLLOW-UP

In both cases, blood glucose levels were in the normal range during outpatient follow-up. Glyburide was effective and was discontinued after 1 mo of oral administration in case 1 and after 1 year in case 2. No major side effects of glyburide were found. The growth and development of the 2 children were similar to that of their peers, and no neurological abnormalities were detected.

DISCUSSION

TNDM is a genetically heterogeneous form of NDM characterized by hyperglycemia and is usually diagnosed before 6 mo of age. It remits during infancy but recurs in later life in most patients[17,18]. Subcutaneous insulin was routinely used to treat NDM previously. However, establishing an effective long-term insulin therapy for NDM is a great challenge for pediatricians and parents because of the irregular feeding habits of infants. Consequently, oral SU therapy has been found to improve glycemic control and to be a more effective in improving the quality of life[19,20].

KATP channels are hetero-octameric complexes formed by four pore-forming Kir6.2 subunits and four SU receptor-1 (SUR1) regulatory subunits and encoded by the KCNJ11 and ABCC8 genes, respectively. In normal pancreatic beta cells, increased glucose across glucose transporter 2 is metabolized by the enzyme glucokinase, resulting in increased production of ATP. This closes the KATP channel, which in turn depolarizes the cell membrane and activates an influx of calcium through voltage-gated calcium channels that subsequently permit exocytosis of insulin granules. ABCC8 gene mutations cause the KATP channels to remain inappropriately open, even in the presence of hyperglycemia. Without channel closure, the cell membrane depolarizes and blocks insulin release from the beta cells, resulting in the clinical manifestations of diabetes mellitus. SU closes the KATP channel through an ATP-independent route, leading to increased insulin secretion[14,21]. Patients with NDM carrying ABCC8 variations have been successfully switched from insulin to oral SU treatment[20,22,23]. However, the appropriate treatment may differ owing to different types of variants and variable clinical phenotypes. In addition, the sensitivity to SU varied among patients with ABCC8 variants. Notably, most
patients, but not all, were successful in transitioning from insulin therapy to SU[22]. The ABCC8 gene, encoding a 1582 amino acid protein, is located on the short arm of chromosome 11 (11p15.1) and comprises 39 exons. Over 700 pathogenic or likely pathogenic ABCC8 mutations have been identified in the ABCC gene, and these include point, missense, nonsense, frame, splicing, and deletion variations[24-26].

A large prospective cohort study by Busiah et al[27] of neonatal diabetes diagnosed before 1 year of age in 68 French centers reported that of 31 patients with NDM caused by an ABCC8 mutation, 11 (13%) were < 1 mo, 19 (27%) were between 1 mo and 6 mo, and 1 (6%) was between 6 mo and 1 year of age. At the end of follow-up, 24 cases were identified as TNDM (78%) and 5 as PNDM (16%)[27]. In 2020, De Franco et al[24] described a total of 748 ABCC8 pathogenic and likely pathogenic variants associated with NDM and congenital hyperinsulinism that had been identified in various countries. To date, the only patients with ABCC8 variants associated with TNDM in China were reported by Li et al[28]. We searched the literature in PubMed and Chinese literature databases, such as China National Knowledge Infrastructure and Wanfang. The ABCC8 gene variants associated with TNDM were reviewed and are summarized in Figure 4, with the majority of variants located in the coding region of the gene. A total of 12 cases with detailed clinical information and the outcomes of SU therapy transfer are summarized in Table 1. The c.3880C>T variation is in the TMD2 domain of the SUR1 subunit in exon 32 of ABCC8 and results in a change from amino acid L to amino acid F at the 1294 position in the encoded protein. According to the ACMG guidelines for classifying genetic variants, this variant is likely to be pathogenic (PM1 + PM2 + PP3 + PP4). So far, no reports of the ABCC8 c.3880C>T variation have been reported, and further functional studies are needed. The age at onset in case 1 was 2 mo, and that in case 2 was 7 mo and 20 d. Both

### Table 1 Clinical features of transient neonatal diabetes mellitus patients with ABCC8 variants (including 2 patients in this case report)

<table>
<thead>
<tr>
<th>Case</th>
<th>Age</th>
<th>DKA</th>
<th>PH</th>
<th>HCO₃⁻</th>
<th>BE</th>
<th>HbA1c</th>
<th>C-peptide</th>
<th>Neurological symptoms</th>
<th>Mutation</th>
<th>Zygosity</th>
<th>Inheritance</th>
<th>Treatment</th>
<th>Ref.</th>
</tr>
</thead>
<tbody>
<tr>
<td>1</td>
<td>36 d</td>
<td>Severe</td>
<td>7.08</td>
<td>3.3</td>
<td>~26</td>
<td>7.6</td>
<td>0.17</td>
<td>No</td>
<td>Arg183Trp</td>
<td>Het</td>
<td>Paternal</td>
<td>Insulin → SU remission in 24 mo</td>
<td>Ngoc et al[29], 2021</td>
</tr>
<tr>
<td>2</td>
<td>50 d</td>
<td>Moderate</td>
<td>7.19</td>
<td>8</td>
<td>~19</td>
<td>7.58</td>
<td>0.41</td>
<td>Convulsion</td>
<td>Gla141Gly</td>
<td>Het</td>
<td>Paternal</td>
<td>Insulin → SU remission in 6 mo</td>
<td>Ngoc et al[29], 2021</td>
</tr>
<tr>
<td>3</td>
<td>35 d</td>
<td>Yes</td>
<td>NA</td>
<td>NA</td>
<td>NA</td>
<td>NA</td>
<td>NA</td>
<td>No</td>
<td>Arg216Cys</td>
<td>Hom</td>
<td>Both parents</td>
<td>Insulin → SU remission in 14 mo</td>
<td>Nayak et al[30], 2021</td>
</tr>
<tr>
<td>4</td>
<td>270 d</td>
<td>No</td>
<td>NA</td>
<td>NA</td>
<td>NA</td>
<td>NA</td>
<td>NA</td>
<td>No</td>
<td>Leu1295Phe</td>
<td>Het</td>
<td>Paternal</td>
<td>Insulin remission in 48 mo</td>
<td>Nayak et al[30], 2021</td>
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<tr>
<td>5</td>
<td>4 d</td>
<td>No</td>
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<td>NA</td>
<td>NA</td>
<td>0.06</td>
<td>NA</td>
<td>NA</td>
<td>Arg1380Cys</td>
<td>NA</td>
<td>NA</td>
<td>Insulin remission in 4 mo</td>
<td>Torbjörnsdotter et al[31], 2020</td>
</tr>
<tr>
<td>6</td>
<td>96 d</td>
<td>NA</td>
<td>NA</td>
<td>NA</td>
<td>NA</td>
<td>7.2</td>
<td>NA</td>
<td>Development delay</td>
<td>Arg653Gln</td>
<td>Het</td>
<td>NA</td>
<td>NA</td>
<td>Balamurugan et al[32], 2019</td>
</tr>
<tr>
<td>7</td>
<td>12 d</td>
<td>NA</td>
<td>NA</td>
<td>NA</td>
<td>NA</td>
<td>0.33</td>
<td>NA</td>
<td>NA</td>
<td>Ile395Phe</td>
<td>NA</td>
<td>NA</td>
<td>Insulin → SU unresponsive remission in 5.5 mo</td>
<td>Li et al[28], 2018</td>
</tr>
<tr>
<td>8</td>
<td>60 d</td>
<td>NA</td>
<td>NA</td>
<td>NA</td>
<td>NA</td>
<td>7.2</td>
<td>&lt; 0.5</td>
<td>NA</td>
<td>Arg877Gln</td>
<td>NA</td>
<td>NA</td>
<td>Insulin remission in 4 mo</td>
<td>Li et al[28], 2018</td>
</tr>
<tr>
<td>9</td>
<td>105 d</td>
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<td>NA</td>
<td>NA</td>
<td>NA</td>
<td>7.9</td>
<td>0.42</td>
<td>NA</td>
<td>Gly1255Ser</td>
<td>NA</td>
<td>NA</td>
<td>Insulin remission in 3.5 mo relapse in 8 yr</td>
<td>Li et al[28], 2018</td>
</tr>
<tr>
<td>10</td>
<td>35 d</td>
<td>No</td>
<td>7.459</td>
<td>22.5</td>
<td>~0.4</td>
<td>4.3</td>
<td>0.6</td>
<td>No</td>
<td>Gly832Cys</td>
<td>Het</td>
<td>De novo</td>
<td>Insulin → SU remission in 1 y and 10 mo</td>
<td>Yamazaki M et al[33], 2017</td>
</tr>
<tr>
<td>11</td>
<td>74 d</td>
<td>Severe</td>
<td>7.06</td>
<td>3.4</td>
<td>~24.8</td>
<td>0.88</td>
<td>9.52</td>
<td>No</td>
<td>Leu1294Phe</td>
<td>Het</td>
<td>De novo</td>
<td>Insulin → SU remission in 1 mo</td>
<td>This study</td>
</tr>
<tr>
<td>12</td>
<td>230 d</td>
<td>Severe</td>
<td>7.12</td>
<td>2.1</td>
<td>~27</td>
<td>1.44</td>
<td>12.63</td>
<td>No</td>
<td>Leu1294Phe</td>
<td>Het</td>
<td>De novo</td>
<td>Insulin → SU remission in 12 mo</td>
<td>This study</td>
</tr>
</tbody>
</table>

HbA1c: Glycosylated hemoglobin; NA: Not available; SU: Sulfonylurea.
patients had ketoacidosis and no family history of diabetes mellitus. During the follow-up period, glyburide proved to be effective and was discontinued after 1 mo and 1 year of oral administration, respectively. No significant side effects of glyburide were observed, suggesting that the age at onset and duration of glyburide treatment differ in patients with the same mutation site. Owing to the ABCC8 variations, patients with TNDM have a high possibility of recurrence in the future, and we will continue to follow-up the children for an extended period.

CONCLUSION

NDM is rarely encountered in clinical practice. In this study, we retrospectively analyzed the clinical phenotypes and ABCC8 c.3880C>T mutations in 2 infants with TNDM. SU glyburide treatment was effective, and this novel de novo variation expands the pathogenic gene mutation spectrum of NDM. All patients diagnosed with diabetes before the 1 year of age should be referred for genetic testing, regardless of their current age, to identify those cases likely to benefit from SU treatment. However, the sample size was limited; therefore, additional evidence and experience should be accumulated over long follow-up periods.

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FOOTNOTES

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