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Dicarboxylic aciduria as a biochemical finding in a 21-month-old girl with recurrent vomiting: a case report

Acyduria dikarboksylowa jako objaw biochemiczny u 21-miesięcznej dziewczynki z nawracającymi wymiotami: opis przypadku

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Abstract

Vomiting in children is a common yet nonspecific clinical symptom with a broad aetiological spectrum, encompassing both benign conditions and rare metabolic disorders. This paper presents the case of a 21-month-old girl with recurrent episodes of vomiting, in whom dicarboxylic aciduria was ultimately diagnosed after extensive differential evaluation. The most common gastroenterological, allergic, and neurological causes were excluded. Metabolic investigations proved crucial, particularly urinary organic acid analysis using gas chromatography–mass spectrometry, which demonstrated the presence of dicarboxylic acids. The case highlights the importance of a broad differential diagnostic approach in children with recurrent vomiting. Early recognition of metabolic disorders enables appropriate patient monitoring and helps prevent potential complications.

Keywords: recurrent vomiting in children, cyclic vomiting syndrome, paediatric inborn defects of metabolism, dicarboxylic aciduria

Streszczenie

Wymioty u dzieci stanowią częsty, lecz nieswoisty objaw kliniczny o szerokim spektrum etiologicznym, obejmującym zarówno łagodne schorzenia, jak i rzadkie choroby metaboliczne. W niniejszej pracy przedstawiono przypadek 21-miesięcznej dziewczynki z nawracającymi epizodami wymiotów, u której po rozległej diagnostyce różnicowej ostatecznie stwierdzono acydurię dikarboksylową. Wykluczono najczęstsze przyczyny gastroenterologiczne, w tym infekcyjne, alergiczne oraz neurologiczne. W diagnostyce kluczowe okazały się badania metaboliczne, w tym analiza kwasów organicznych w moczu metodą chromatografii gazowej/spektrometrii mas, które wykazały obecność kwasów dikarboksylowych. Przedstawiony przypadek podkreśla znaczenie szerokiej diagnostyki różnicowej u dzieci z nawracającymi wymiotami. Wczesne rozpoznanie zaburzeń metabolicznych umożliwia właściwe monitorowanie pacjenta i zapobieganie potencjalnym powikłaniom.

Słowa kluczowe: nawracające wymioty u dzieci, zespół cyklicznych wymiotów, pediatryczne wady metabolizmu, acyduria dikarboksylowa

INTRODUCTION

Vomiting is one of the most common reasons why caregivers seek medical attention for children, and its aetiology is diverse. It may reflect mild, self-limiting disorders, but it can also be the first manifestation of severe, potentially life-threatening systemic disease. Most commonly, vomiting is associated with gastrointestinal infections, particularly rotavirus infection, which typically presents with diarrhoea and vomiting, and may lead to dehydration[1,2].

Another relatively common cause of vomiting is gastro-oesophageal reflux. In infants, it is often physiological. It usually appears before the 8th week of life and resolves before the end of the first year. The backflow of gastric contents into the oesophagus leads to regurgitation. Although this causes considerable parental concern, it usually does not require extended diagnostic evaluation or pharmacological treatment. Nevertheless, diagnostic vigilance should be maintained, as physiological reflux in infants may eventually progress to gastro-oesophageal reflux disease, resulting in oesophagitis and acquired oesophageal stricture[3]. Further diagnostic evaluation is necessary when alarm symptoms occur, such as the presence of blood or bile in the vomitus or projectile vomiting[3].

Vomiting accompanied by diarrhoea, abdominal distension, and abdominal pain, together with growth retardation, delayed puberty, chronic fatigue, or anaemia, should prompt the exclusion of autoimmune diseases such as coeliac disease or eosinophilic esophagitis[4,5]. In the differential diagnosis of vomiting aetiology, food allergy and food protein-induced enterocolitis syndrome (FPIES) should also be considered. FPIES is a severe, non-IgE-mediated gastrointestinal food allergy which, in its acute form, manifests as intense, recurrent vomiting, often accompanied by diarrhoea, leading to dehydration and pathological lethargy. It may present in either an acute or chronic form, and may affect both children and adults[6]. In the acute form, symptoms usually occur 1 to 4 hours after ingestion of the triggering food. The diagnosis requires fulfilment of the major criterion and at least three minor criteria (Tab. 1)[6]. The chronic form manifests with prolonged or recurrent vomiting and/or diarrhoea, which may lead to systemic complications such as failure to gain weight, dehydration, hypoalbuminaemia, or acidosis[6].

In cases of recurrent vomiting, cyclic vomiting syndrome (CVS) should also be included in the differential diagnosis. This syndrome is characterised as a disorder of gut-brain interaction[7]. The diagnosis of CVS is based on the Rome V criteria, according to which the diagnosis requires the occurrence of two or more periods of intense, paroxysmal, stereotypical vomiting lasting no longer than seven days, occurring at least twice within the previous six months, and separated by periods of normal health[8]. CVS is estimated to affect approximately 2% of the population in the United States, 1% of the population in the United Kingdom, and 0.7% of the population in Canada[9].

FPIES subtype	Diagnostic criteria	Clinical presentation
Acute	**Major criterion:** vomiting within 1–4 hours after ingestion of the suspected food, without typical symptoms of IgE-mediated allergy **Minor criteria:** recurrent vomiting after ingestion of the same or another food, lethargy, pallor, need for emergency department evaluation, requirement for intravenous fluid therapy, diarrhoea within 24 hours, hypotension, hypothermia Diagnosis requires fulfilment of the major criterion and at least three minor criteria	Acute, recurrent gastrointestinal symptoms following exposure to the triggering food
Chronic	Chronic or intermittent vomiting and/or watery diarrhoea, together with impaired weight gain during regular exposure to the causative food. Confirmation is based on resolution of symptoms after elimination of the food and recurrence after its reintroduction	Possible dehydration, hypoalbuminaemia, acidosis, methaemoglobinaemia, and growth impairment

Tab. 1. Diagnostic criteria for food protein-induced enterocolitis syndrome (FPIES)

The exact pathophysiology of CVS remains unknown. However, it has been associated with environmental, genetic, autonomic, and neurohormonal factors, as well as migraine. It should be emphasised that CVS is a diagnosis of exclusion. Therefore, abdominal imaging and upper gastrointestinal endoscopy should first be performed[7], and other organic and external causes of recurrent vomiting should be ruled out, including psychological stress; gastrointestinal diseases (i.e. eosinophilic esophagitis, coeliac disease, inflammatory bowel disease, peptic ulcer disease of the stomach and duodenum, post-viral gastroparesis, superior mesenteric artery syndrome); infections; motion sickness; allergies; mitochondrial disorders; benign torticollis of infancy; neurological disorders (early benign childhood occipital epilepsy, brain tumours, migraine, tension-type headaches, benign paroxysmal vertigo); and metabolic and endocrinological disorders (inborn errors of metabolism, adrenal insufficiency, type 1 diabetes mellitus, hereditary fructose intolerance)[10,11].

In the differential diagnosis of recurrent vomiting, inherited metabolic diseases should also be excluded. Investigations for organic acidaemias, fatty-acid oxidation disorders, and urea-cycle defects may be helpful.

The present paper describes the case of a 21-month-old girl, who was hospitalised several times because of recurrent vomiting, in whom, after extensive diagnostic evaluation, a rare metabolic disorder – dicarboxylic aciduria – was diagnosed.

CASE REPORT

A 21-month-old girl (G1P1, spontaneous vaginal delivery at 38 weeks' gestation, birth weight 3,220 g, birth length 52 cm, unremarkable perinatal history) was hospitalised several times

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Capillary blood gas parameter	Reference range	Hospitalisation I	Hospitalisation II	Hospitalisation III
pH	7.37–7.45	7.397	7.322	7.346
pCO_2 [mm Hg]	35.0–46.0	30.6	31.8	24.4
pO_2 [mm Hg]	70.0–100.0	63.0	80.0	95.0
HCO_3 [mmol/L]	21.0–26.0	18.8	16.5	13.9
BE [mmol/L]	−2.0–+3.0	−6.0	−9.6	−11.8
Sodium [mmol/L]	135.0–145.0	137.0	143.0	141.0
Potassium [mmol/L]	3.6–4.8	5.7	4.4	4.5
Chlorides [mmol/L]	95.0–105.0	107.0	121.0	108.0
Glucose [mg/dL]	70.0–99.0	107.0	81.0	77.0

Tab. 2. Results of biochemical investigations obtained during subsequent hospitalisations

in the Department of Paediatrics, Paediatric Nephrology and Allergology, Military Institute of Medicine – National Research Institute, due to recurrent episodes of vomiting.

According to her medical history, the girl had experienced recurrent vomiting for approximately one year, usually consisting of 3–4 episodes of emesis containing food contents, with spontaneous resolution. Each episode occurred without other concerning symptoms such as cough, rhinorrhoea, fever, or diarrhoea.

The first hospitalisation in the department occurred when the girl was 18 months old. On admission, her general condition was assessed as moderate. Physical examination revealed a hypotrophic body habitus and scant subcutaneous tissue (body weight at the 3rd percentile, height at the 25th percentile). Diagnostic evaluation was conducted in a stepwise manner, according to the clinical presentation and with the aim of excluding potential causes of the reported symptoms. Initially, basic laboratory tests assessing the child's general condition were performed. Capillary blood gas analysis revealed metabolic acidosis with base deficit (Tab. 2). No abnormalities were found in the complete blood count, and inflammatory markers were low. Subsequently, investigations for infectious causes of the symptoms were performed. Parasitic infestation of the gastrointestinal tract was excluded. Stool tests for rotavirus, adenovirus, and norovirus were negative, as were bacteriological stool cultures, with no growth of *Salmonella*, *Shigella*, or *Yersinia* species.

Due to the persistence of gastrointestinal symptoms, the diagnostic work-up was extended to include investigations for immunological and allergic disorders. Serum immunoglobulin concentrations (IgA, IgG, and IgM) were within normal ranges, as were thyroid hormone levels. Metabolic parameters, including muscle enzyme activity, ammonia concentration (26 μmol/L; reference range 11–51 μmol/L), and glucose levels, were normal. The concentration of anti-tissue transglutaminase antibodies was low, arguing against coeliac disease. No allergen-specific IgE antibodies to the tested inhalant and food allergens were detected.

At a later stage, imaging studies were performed. Abdominal ultrasonography showed no abnormalities of the parenchymal organs. Given the nature of the symptoms and the need to exclude central nervous system causes, an ophthalmologic consultation was obtained; fundoscopy revealed no signs of increased intracranial pressure. Magnetic resonance imaging of the head, performed under general anaesthesia, showed no focal brain lesions. Based on the overall clinical picture and the results of the investigations performed, gastro-oesophageal reflux was suspected. The girl was discharged home with recommendations for anti-reflux medication and follow-up in the department for electroencephalography (EEG).

The patient was rehospitalised in the department one month later because of another vomiting episode. At that time, inflammatory markers remained low, and liver and kidney function parameters were normal. Capillary blood gas analysis again demonstrated features of metabolic acidosis (Tab. 2). No serum water-electrolyte disturbances were found. Ammonia concentration was within the normal range (27 μmol/L; reference range 11–51 μmol/L). An EEG was performed and showed no abnormalities. Stool tests for norovirus, adenovirus, and rotavirus were repeated and again yielded negative results. During hospitalisation, the girl received intravenous fluid therapy. She was discharged home in good general condition.

A decision was made to determine the urinary organic acid profile and acylcarnitine profile directly during the next vomiting episode. This was based on the fact that many inborn errors of metabolism become apparent only during catabolic states. During the interictal period, test results may remain normal or show only nonspecific diagnostic abnormalities. Moreover, these tests should be performed before the administration of supportive intravenous infusions, in order to avoid falsification of the results caused by infusion-related alterations in metabolite profiles.

Another vomiting episode occurred when the girl was 21 months old. On admission, her general condition was moderate. Physical examination findings were similar to those observed during previous hospitalisations. Additional laboratory tests showed mildly elevated inflammatory markers (1.8 mg/dL; normal <0.5 mg/dL). Complete blood count revealed leukocytosis with neutrophilia (18.15×10^9/L, neutrophils 67.4%). Renal and liver function parameters were within age-appropriate reference ranges. Urinalysis showed no signs of urinary tract infection, trace proteinuria (15 mg/dL), and ketonuria (80 mg/dL). Capillary blood gas analysis again demonstrated metabolic acidosis (Tab. 2), without significant electrolyte disturbances.

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Screening for inborn errors of metabolism using tandem mass spectrometry revealed a slightly elevated concentration of C4-OH (3-hydroxybutyrylcarnitine) (0.783 μmol/L; reference value <0.72). The amino acid profile was normal. Urinary organic acid profiling was performed using gas chromatography-mass spectrometry (GC-MS). The sample showed markedly elevated levels of acetoacetic acid and 3-hydroxybutyric acid, as well as elevated levels of 2-hydroxybutyric acid, 2-methylacetoacetic acid, and dodecanedioic acid. In addition, mildly increased levels of 2-methyl-3-hydroxybutyric acid, 3-hydroxyisovaleric acid, 2-ketoisovaleric acid, ethylhydracrylic acid, 2-keto-3-methylvaleric acid, 2-ketoisocaproic acid, hippuric acid, 3-hydroxyoctenedioic acid, 3-hydroxysuberic acid, 3-hydroxysebacic acid, and 3-hydroxydodecanedioic acid were found.

These findings indicated dicarboxylic aciduria with massive ketonuria. The girl was treated with supportive intravenous fluid therapy, resulting in improvement in her general condition. Follow-up capillary blood gas analysis showed no evidence of acidosis. Based on the overall clinical picture and the results of additional investigations, the girl was diagnosed with dicarboxylic aciduria. The child, in good general condition, was subsequently referred to a specialised department for inborn errors of metabolism and is currently awaiting genetic testing.

DISCUSSION

Dicarboxylic aciduria is characterised by markedly increased urinary excretion of dicarboxylic fatty acids. It reflects a metabolic shift in free fatty acid oxidation from mitochondrial β-oxidation to microsomal ω-oxidation, resulting in the formation of α,ω-dicarboxylic acids that are subsequently partially metabolised, conjugated, and excreted in the urine[12,13]. The most common causes are inherited mitochondrial defects of β-oxidation (e.g. medium-chain acyl-CoA dehydrogenase deficiency – MCADD, very-long-chain acyl-CoA dehydrogenase deficiency – VLCAD, long-chain 3-hydroxyacyl-CoA dehydrogenase deficiency/mitochondrial trifunctional protein deficiency – LCHAD/MTP), electron transfer flavoprotein disorders (multiple acyl-CoA dehydrogenase deficiency – MADD) and peroxisomal diseases[12–14]. Dicarboxylic aciduria may also occur secondarily as a result of medium-chain triglycerides (MCT)-rich diets, certain medications, or catabolic states[12–14].

In the present patient, urinary organic acid profiling was performed using GC-MS. The results demonstrated marked ketonuria and elevated urinary levels of dicarboxylic acids, including 3-hydroxyoctenedioic, 3-hydroxysuberic, 3-hydroxysebacic, and 3-hydroxydodecanedioic acids. These abnormalities suggested a disorder of fatty acid oxidation.

Proper collection of blood and urine is essential to ensure analytical quality and the reliability of metabolic test results. For dried blood spot sampling, capillary blood is obtained by puncturing the lateral surface of the fingertip (third or fourth finger of the non-dominant hand) after skin disinfection and drying. The first drop should be discarded; subsequent drops should flow freely without squeezing the finger and be applied so that each filter-paper circle is evenly saturated and visibly soaked through to the reverse side. Cards should be dried flat at room temperature for 2–4 hours away from light, heat sources and humidity, and drying must not be accelerated. Once fully dry, each card should be placed in an individual air-permeable envelope with a desiccant and humidity indicator, avoiding sealed plastic packaging. Transport conditions should minimise exposure to high temperature and humidity; for particularly labile assays temperature control at 2–8°C and the shortest possible transit time, in accordance with the reference laboratory's recommendations, are advised. For urine GC-MS analysis, first-morning midstream urine is preferred (sterile collection bags for infants); the sample should be cooled and delivered promptly, and, if delay is unavoidable, stored at ≤–20°C. Improper drying, storage, or transport may cause analyte degradation, altered concentrations, and false-positive or false-negative results[15].

As noted above, MADD should be considered in the differential diagnosis of dicarboxylic aciduria[14]. MADD is an autosomal recessive disorder of the ETF–ETFDH (electron transfer flavoprotein–ETF dehydrogenase) complex (ETFA, ETFB, ETFDH), resulting in impaired oxidation of fatty acids and certain amino acids, with accumulation of acyl-CoA species, acylcarnitines, and various organic acids. The clinical spectrum ranges from severe neonatal forms with metabolic acidosis, profound hypoglycaemia, hyperammonaemia, hepatomegaly, and congenital anomalies (types I–II) to late-onset forms (type III), presenting from childhood to adulthood with chronic myopathy, exercise intolerance, myalgias, episodes of rhabdomyolysis, and intermittent metabolic decompensations[14]. Diagnosis combines biochemical and genetic testing. A hallmark finding is multi-chain elevation of acylcarnitines on MS/MS together with numerous organic acids in urine (glutaric and ethylmalonic acids, dicarboxylic and 3-hydroxy acids, acylglycines). Assessment of carnitine, glucose, ammonia, creatine kinase, and liver function is also required. Definitive confirmation is obtained by identifying biallelic pathogenic variants in ETFA/ETFB/ETFDH or by enzymatic assays in fibroblasts. In routine care, prevention of crises is essential: avoiding prolonged fasting, ensuring regular feeding, and having a clear sick-day plan so caregivers know when to give oral carbohydrates and when urgent hospital assessment is required. In acute crises, the priority is rapid cessation of catabolism with high-dose carbohydrates (often intravenous glucose), fat restriction, and correction of metabolic acidosis; severe hyperammonaemia may require haemodialysis. Long-term management focuses on crisis prevention through frequent meals, a carbohydrate-rich, fat- and protein-restricted diet, along with chronic supplementation with riboflavin, L-carnitine, and coenzyme Q10. In the present case, episodic

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metabolic acidosis with vomiting, the urinary organic acid profile (multiple 3-hydroxy acids, dicarboxylic acids and other amino-acid-related metabolites), and a slight elevation of C4–OH are compatible with a multisystem acyl-oxidation defect such as MADD, whereas massive ketonuria together with normal plasma ammonia and glucose concentrations argues against it; therefore, genetic testing is required to establish the definitive diagnosis[14].

Another group of disorders to consider comprises inherited mitochondrial defects of β-oxidation, including MCADD. MCADD is an autosomal recessive defect of mitochondrial β-oxidation of medium-chain fatty acids, most commonly caused by pathogenic variants in ACADM (acyl-CoA dehydrogenase medium chain), classically c.985A > G). This results in loss of enzymatic activity, accumulation of acyl-CoA species and acylcarnitines, and a metabolic shift towards ω-oxidation with consequent urinary excretion of dicarboxylic acids[16].

Clinically, MCADD typically presents in infancy or during catabolic stress (fasting or intercurrent infection) with acute episodes characterised by hypoketotic hypoglycaemia, vomiting, anorexia, apathy/lethargy, seizures, loss of consciousness, and potentially coma. More severe presentations may include hepatomegaly, elevated transaminases, metabolic acidosis, increased creatine kinase, and hyperammonaemia. In some patients, the first manifestation may be sudden death. Cardiac manifestations (QT prolongation, tachyarrhythmias, rarely cardiomyopathy), as well as later or milder phenotypes (fatigability, muscle weakness, reduced exercise tolerance, and rhabdomyolysis) have also been described. Long-term neurological sequelae (developmental delay, loss of acquired skills, attention deficits) may persist after decompensation; in some individuals, the disease remains asymptomatic until the first metabolic crisis[16].

Acute management of an MCADD crisis focuses on immediate cessation of catabolism. If oral glucose administration is not feasible, an initial bolus of 25% dextrose at 2 mL/kg should be administered, followed by continuous infusion of 10% dextrose at 1.5 times the maintenance rate until blood glucose concentrations ≥5 mmol/L are achieved, with blood glucose monitoring every 15–30 minutes and close surveillance of electrolytes, blood gases, liver transaminases, and creatine kinase activity. Preventive care requires parental education, avoidance of fasting, and regular feeding. L-carnitine supplementation may be considered, although its benefit remains uncertain. Newborn screening has markedly improved outcomes in MCADD, but rapid recognition and management of decompensation remain crucial determinants of prognosis[16].

In the present patient, the GC-MS profile is poorly characteristic of MCADD because of the prominent ketonuria, whereas classical MCADD is associated with hypoketotic hypoglycaemia. The presence of ethylmalonic acid and a broad spectrum of 3-hydroxy dicarboxylic acids favours secondary activation of ω-oxidation or a generalised β-oxidation defect rather than isolated ACADM deficiency. Furthermore, the absence of elevated transaminases and hypoglycaemia in this patient argues against MCADD.

In the differential diagnosis of dicarboxylic aciduria, secondary causes must be considered. One such cause is excessive intake of medium-chain fatty acids (MCFAs), which can overwhelm mitochondrial β-oxidation. Pharmacologic inhibition of mitochondrial pathways and catabolic states that increase lipolysis can likewise shift fatty-acid oxidation towards microsomal ω-oxidation, resulting in the formation and urinary excretion of dicarboxylic acids (C6–C12) and their 3-hydroxy derivatives[16,17].

Dietary supplementation with medium-chain triglycerides (e.g. in infant formulas or MCT oils) increases MCFA load and has been associated with increased urinary excretion of dicarboxylates and acylglycines, thereby mimicking the biochemical phenotype of primary β-oxidation defects[16,17]. Certain drugs, including valproic-acid derivatives and dipropylacetate, may also affect mitochondrial metabolism and ω-oxidation, leading to elevation of C6–C10 dicarboxylic acids in urine[18].

Finally, periods of fasting, intercurrent infection, or other catabolic stress amplify lipolysis, increase the substrate load for β-oxidation, and precipitate mitochondrial overload, which can likewise cause secondary dicarboxylic aciduria. In such cases, the biochemical profile often normalises after resolution of the catabolic state[16]. Clinically, distinguishing secondary dicarboxylic aciduria relies on correlating the biochemical pattern with the history (MCT exposure, medications, fasting) and on test reproducibility: secondary forms typically resolve after removal of the triggering factor or restoration of an anabolic state, whereas primary β-oxidation defects are characterised by a reproducible, specific acylcarnitine pattern and warrant further genetic evaluation[16,17].

Our patient requires further evaluation at a specialised metabolic centre for primary disorders of acyl oxidation, including urgent panel genetic testing (ETFA, ETFB, ETFDH, ACADM, and other βoxidation genes). Biochemical investigations should be repeated and expanded – acylcarnitine profiling and measurement of total and free carnitine during an acute episode and in the interictal period, repeat urinary organic acid analysis (GCMS), and serial measurements of glucose, ammonia, liver transaminases, and creatine kinase. These tests should be performed during subsequent decompensation episodes and in the intercritical state to confirm reproducibility of the biochemical pattern or to exclude a secondary cause.

CONCLUSIONS

Recurrent vomiting in children necessitates not only exclusion of common gastrointestinal causes but also consideration of metabolic, neurological, and systemic disorders. Basic metabolic investigations (acylcarnitine profile, plasma and urine organic acids, glucose, ammonia, and carnitine)

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should be obtained during an acute episode and repeated in the intercritical period, as only this approach allows differentiation of transient biochemical perturbations from true inborn metabolic defects. Abnormal results warrant prompt consultation with a metabolic centre and genetic testing to establish a definitive diagnosis.

In routine practice, priority should be given to preventing and rapidly treating decompensation episodes – regular feeding, avoidance of prolonged fasting, clear sick-day instructions for caregivers, and early provision of carbohydrates with supportive therapy during crises. Long-term outcomes are improved by active collaboration with caregivers and multidisciplinary care, which together facilitate clinical decision-making and optimise prognosis.

Conflict of interest

The authors declare no conflicts of interest.

Author contribution

Original concept of study: NG, AT, AZ. Collection, recording and/or compilation of data: NG, AT, AZ, AJ. Analysis and interpretation of data: NG, AT, AZ, AB, JJ, SMJ, KP. Writing of manuscript: NG, AB, JJ, AJ, SMJ, KP, AR. Critical review of manuscript: AR. Final approval of manuscript: NG.

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