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CASE BASED REVIEW

Can hyperuricemia predict glycogen storage disease (McArdle's disease) in rheumatology practice? (Myogenic hyperuricemia)

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Abstract

Gout disease is an inflammatory arthritis that arises due to the accumulation of monosodium urate crystals (MSU) around the joints and in tissues. Clinical manifestation of metabolic diseases leading to secondary hyperuricemia most predominantly occurs in the form of gouty arthritis. Hyperuricemia and gout may develop during the course of glycogen storage diseases (GSD), particularly in GSD type I, which involves the liver. On the other hand, during the course of GSD type V (GSDV, McArdle's disease), which merely affects the muscle tissue due to the deficiency of the enzyme myophosphorylase, hyperuricemia and/or gout is rarely an expected symptom. These patients may mistakenly be diagnosed as having idiopathic hyperuricemia and associated gout, leading to the underlying secondary causes be overlooked and thus, diagnostic delays may occur. In this case report, we present a premenopausal female patient who experienced flare-ups of chronic arthritis while on disease-modifying antirheumatic drugs and intraarticular steroids due to a diagnosis of undifferentiated arthritis. The patient was initially suspected of having gouty arthritis because elevated concentrations of uric acid were incidentally detected, but then, a diagnosis of asymptomatic GSDV was made owing to elevated concentrations of muscle enzymes during colchicine use. Our aims were to remind rheumatologists of the phenomenon of "myogenic hyperuricemia" and to discuss the potential causes of hyperuricemia that develop during GSD along with the available literature.

Keywords Glycogen storage disease type 5 · Gout · Hyperuricemia · McArdle's disease

Introduction

Gout is an autoinflammatory disease characterized by hyperuricemia, recurrent arthritis flare-ups, and storage of monosodium urate crystals in the joints and peripheral tissues. Gout is more common among males and usually develops around the fifth decade of life. Due to the uricosuric effects of estrogen, the prevalence of gout in females is very low in the premenopausal period [1].

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Glycogen storage disease (GSD) is a metabolic disease that affects the liver, skeletal muscle, heart, and occasionally, central nervous system and kidneys, which occurs as a result of the deficiencies of enzymes acting on glycogen synthesis, glycogen catabolism, or glycolysis. In GSD type I, which can particularly involve the liver and/or muscle, in addition to causing metabolic disorders such as hypoglycemia and lactic acidosis, hyperuricemia and gout are well-defined, whereas in GSD type V (GSDV, McArdle's disease), which is the deficiency of the enzyme muscle phosphorylase, hyperuricemia and/or gout are very seldom reported [2, 3]. Patients with various forms of GSD, particularly those with type I, are known to have hyperuricemia that arises due to the increased degradation of adenosine triphosphate (ATP) as a result of metabolic liver failure [4]. In GSDV, hyperuricemia was attributed to the increase in purine metabolites due to the accelerated catabolism of purine nucleotides following muscle exercise [5, 6].

Our aim with this case report is to illustrate that GSD may occur even in adult patients and should be considered in the differential diagnosis of inflammatory myopathies and that the

presence of hyperuricemia in patients with myopathy can indicate McArdle's disease. Moreover, we wanted to remind rheumatologists of the 'myogenic hyperuricemia' phenomenon and discuss the potential causes of hyperuricemia that develop during GSD.

Search strategy

For this purpose, we searched MEDLINE, Web of Science, and Scopus databases using MeSH terms *Glycogen storage disease type V* [AND] *gout*, *Glycogen storage disease type V* [AND] *hyperuricemia*, *McArdle's disease* [AND] *gout*, and *McArdle's disease* [AND] *hyperuricemia* up until February 20, 2019. Case series and case reports were included in the study. Reviews and publications on the relationship between non-type-V GSDs and gout/hyperuricemia and articles not written in English were excluded from the study. A flowchart depicting the search strategy of our study is shown in Fig. 1. Altogether, six articles plus our case (total eight patients) were analyzed [2, 3, 5–8]. In order to make a clearer demonstration of the relationship between elevated uric acid levels and GSD, the data of our five age- and sex-matched patients with polymyositis (PM) were compared with the data of our patient with gout.

Case presentation

A 35-year-old female patient, who first presented in 2007 aged 24 years, presented with pain and swelling in bilateral knees and her left ankle. In her physical examination, arthritis was detected in her left subtalar region, right ankle, and left knee, but she had no nail changes, skin-related signs, or enthesitis to suggest psoriasis. Undifferentiated arthritis (psoriatic arthritis) was considered because her mother had a history of psoriasis, and treatment with sulfasalazine and oral corticosteroid was initiated and intraarticular steroid injection was performed [erythrocyte sedimentation rate (ESR): 56 mm/h, C-reactive protein (CRP): 4.57 mg/dL (normal range: 0–0.5 mg/dL)]. An HLA B27 test was negative and the patient had no sacroiliitis.

She was lost to follow-up for 2 years. Arthritis in the 2nd metacarpophalangeal joint of the right hand, right subtalar arthralgia, and minimal fluid in the left knee was detected in 2009, and treatment with non-steroid anti-inflammatory drug was initiated. Sulfasalazine and methotrexate were used due to the progression of the arthritis in the patient's left knee. Due to

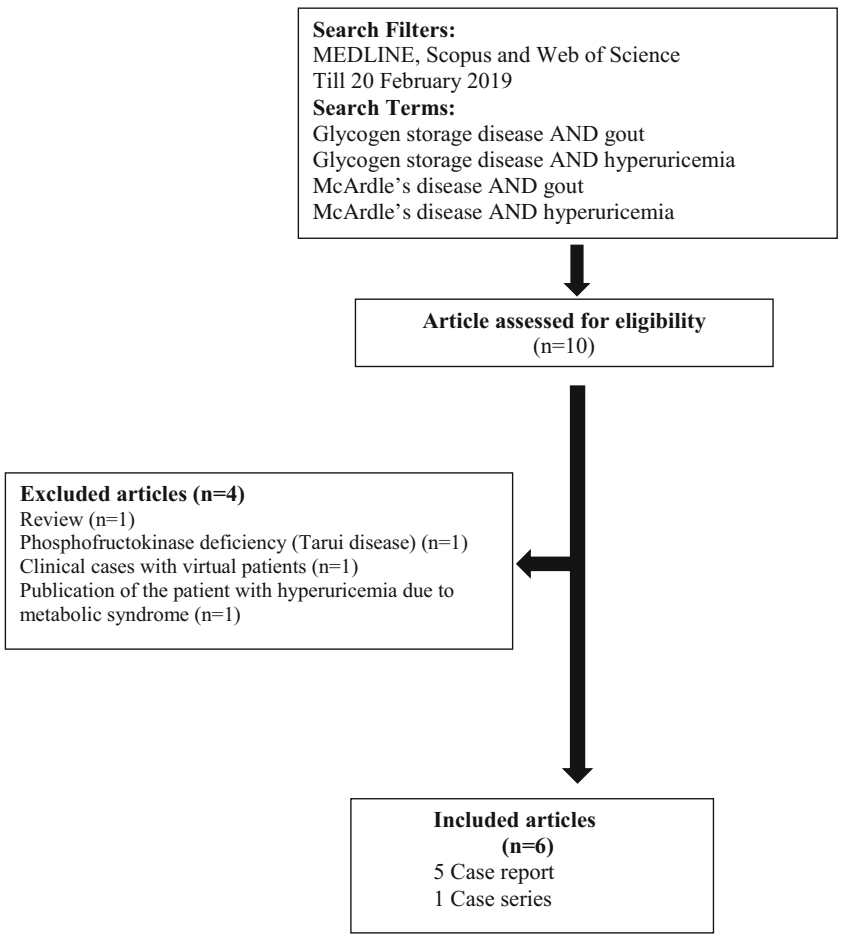

Fig. 1 Flowchart of the search strategy of the review

unresponsiveness, treatment with golimumab was initiated in February 2015 in combination with isoniazid prophylaxis because of a positive tuberculin test. However, as an alternative diagnosis, the possibility of gout was considered because of hyperuricemia [uric acid concentration: 13.1 mg/dL (normal range: 2.4–5.7 mg/dL; uricase-based enzymatic colorimetry)]. The presence of monosodium urate (MSU) crystals could not be tested because there were no problematic joints from which synovial samples could be collected during this period. The patient's joint problems, which had continued for the last 10 years, were re-evaluated. The patient stated that the joint problems she experienced during her first admission in 2007 were in the form of attacks that occurred twice a year and entered into remission within 10 days. When the patient was re-examined in detail, a small tophus was detected at the dorsum of her hand. When the patient was questioned for potential causes of hyperuricemia, there were no drug use, hypertension and renal insufficiency history, or a family history of gout. She had a normal body mass index (height: 156 cm, weight: 53 kg, body mass index 22.6 kg/m^2). Measurement of hypoxanthine phosphoribosyl concentrations was planned transferase to reveal any enzyme deficiency suggesting primary gout because the patient was a young premenopausal female. Owing to laboratory-related problems, xanthine and hypoxanthine concentrations were measured in urine instead of the enzyme, and both were found to be low [xanthine: 0.15 nmol/L (normal range: 0.33–0.85 nmol/L), hypoxanthine: 0.01 mmol/L (normal range: 10.3–11.1 nmol/L)]. Upon the diagnosis of gout, golimumab was discontinued; treatment with allopurinol 150 mg/day and colchicine was initiated. Febuxostat 80 mg/day was used instead of allopurinol because the latter caused skin rash (uric acid concentration at month 3 of allopurinol: 10.4 mg/dL).

When the patient was checked for any adverse effects of colchicine, her creatine kinase (CK) concentration was 410 IU/L and colchicine dose was reduced. The CK concentration then decreased to 291 IU/L. In the ongoing process, as CK increased to 1013 IU/L and decreased to 451 IU/L after colchicine was discontinued, the treatment was changed to the interleukin-1 receptor antagonist, anakinra. Meanwhile, the patient had arthritis again in the interim period between colchicine discontinuation and initiation of anakinra; MSU crystals were observed in synovial fluid. While on febuxostat (uric acid level at month 3 of treatment: 4.9 mg/dL) and anakinra treatment, the patient's CK concentrations ranged between 1293 and 2132 IU/L. Muscle damage due to the possible intramuscular injection of anakinra was suspected, and the treatment was discontinued.

The muscle strength of the patient was evaluated in the bilateral upper and lower extremities according to the Medical Research Council (MRC) Scale for Muscle Strength, which resulted as normal. As a pre-diagnosis of myopathy was established based on CK levels around 1200–2000 IU/L during the follow-ups, electroneuromyography was conducted, which resulted as normal. No inflammatory cell infiltration was observed in the muscle biopsy, glycogen accumulation was detected in periodic acid–Schiff (PAS) staining, enzyme histochemical and immunohistochemical analysis for phosphorylase was negative, and phosphofructokinase was positive (Figs. 2 and 3). McArdle's disease was confirmed at the molecular level with supporting results of enzyme concentrations and genetic analysis. The patient's results are presented in detail in Table 1. The patient was accepted as having GSD (McArdle's disease)-induced myopathy.

Retrospective questioning of the patient revealed that she had exercise intolerance present from childhood and had 'second wind' phenomenon, which is a pathognomonic feature of McArdle disease [the second wind denotes a sudden, marked improvement in the tolerance to aerobic, dynamic, large muscle mass exercise (walking or cycling) after about 10 min, that is, disappearance of the excessive fatigue, breathlessness, and tachycardia that were triggered by the start of exertion]. Most patients describe this phenomenon as the ability to resume exercising (e.g., walking or brisk walking in the more-fit patients) if they take a brief rest at the appearance of premature fatigue early during exercise [7, 9]).

The patient's follow-up on febuxostat treatment is ongoing. She was instructed to stay away from strenuous exercise because of GSDV and to consume a carbohydrate-rich diet before exercising if she was going to do intense exercise (latest tests: ESR: 24 mm/h, CRP: 0.331 mg/dL, uric acid: 5.9 mg/dL, CK: 1502 IU/L, LDH: 239 IU/L).

Discussion

Gout is predominantly encountered in males and postmenopausal females. Gout that develops at an early age usually

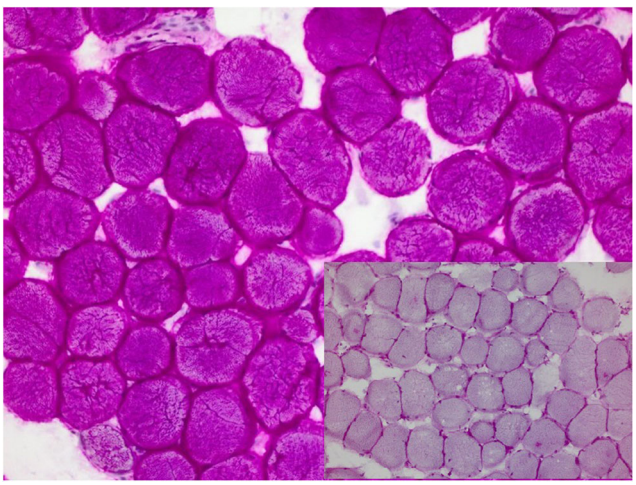

Fig. 2 PAS shows subsarcolemmal intensification at some fibers and an overall increased cytoplasmic staining that is washed out with diastase (lower right) (× 20)

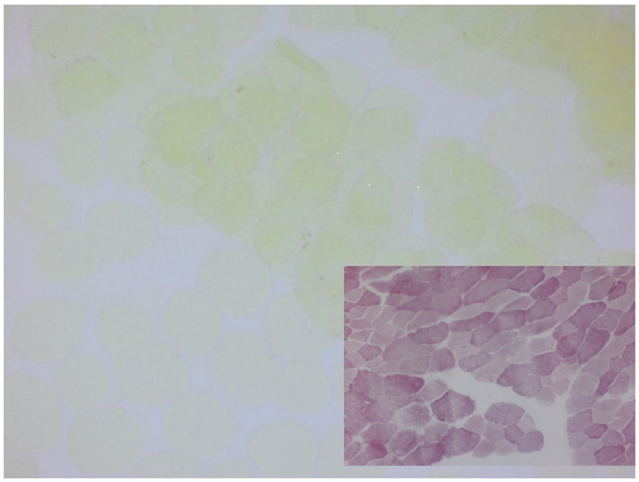

Fig. 3 Myophosphorylase histochemical staining along with a positive control (lower right) shows no staining of myofibers (× 20)

indicates an underlying genetic disease. Genetic disorders of purine metabolism must be investigated in young premenopausal female patients with gout who have no hypertension or renal insufficiency [10]. Considering that our patient was young, premenopausal, and had gout, we searched for any purine metabolism disorder, and we concluded that she had GSD after she experienced asymptomatic myopathy.

GSD is a class of inherited genetic disorders that cause glycogen storage as a result of a lack of glycolysis due to an enzyme deficiency at a certain step. GSD primarily affects liver and muscle tissue, as well as the organs such as heart and spleen. Von Gierke disease (GSD type I, hepatorenal glycogenosis) is an autosomal recessive disorder characterized by the failure to convert the stored hepatic glycogen into circulating glucose. GSD type I may manifest itself with glycogen deposits in the liver and other tissues, hepatomegaly, in particular, as well as hypoglycemic flares, hypertriglyceridemia, lactic acidosis, hyperuricemia, and developmental and mental retardation. Gout may develop in affected children and adults [11]. Gout (hyperuricemia) has been well defined in inherited GSDs, particularly in the course of GSD type I. In a study including a total of 17 patients with GSD type I with gout and tophus, 10 patients (62.5%) were diagnosed as having GSD after the diagnosis gout was made [12].

Other than GSD type I, hyperuricemia/gout is also reported in GSD types III and VII, albeit rarely [6, 13]. The occurrence of gout/hyperuricemia in GSDV is very seldom [2, 3, 5–8]. Type V is an autosomal recessive trait and caused by muscle glycogen phosphorylase (myophosphorylase) deficiency [11]. Overall, it is difficult to make a clear and definitive estimation of the ratio for hyperuricemia in GSD; however, it was reported to be approximately 50% [14]. We only found six publications in the literature that included gout and/or hyperuricemia together with GSDV. In total, eight patients including ours were analyzed. Gout was described in four patients (our patient was the 4th patient in the literature), and hyperuricemia alone was described in four cases. Although all patients with gout were aged over 45 years, our patient was aged 35 years; seven of the eight patients were male, only our patient was female and she was the first female patient in the literature to be diagnosed as having GSDV + gout. Similar to the study by Zhang B et al., patients were diagnosed as having GSD after the diagnosis of gout was made [12]. There is usually a delay in diagnosing McArdle's disease. In a group comprising 50 patients with a mean age of 48 years, the median duration from false diagnosis to correct diagnosis was 23 years (interval: 1–62 years) [7]. Out of the patients discussed in this study, three of four patients diagnosed as having GSDV/gout were diagnosed as having GSD 10, 23, and 24 years after the initial joint symptoms. The other patient was diagnosed as having GSD and gout simultaneously because the attack symptoms were ongoing for 6 years. In terms of GSD-related myopathy, only six of the eight patients had muscle symptoms, whereas the remaining two patients were diagnosed as having myopathy upon determination of asymptomatic elevated CK concentrations while on colchicine treatment. Examination of muscle biopsy samples revealed enzyme deficiency in six patients and glycogen accumulation in three patients. The deficiency of myophosphorylase enzyme was biochemically demonstrated in five patients, whereas the mutation for GSDV (PYGM mutation) was demonstrated in only two patients [2, 3, 5–8]. The demographic, clinical, and laboratory characteristics of our patient and the patients in the literature, including the GSDV and gout/hyperuricemia cases, are given in Table 1.

Why does hyperuricemia occur in patients with GSD? There are certain theories about the pathogenesis of hyperuricemia occurring during the course of GSD. It was suggested that in GSD type I, both uric acid production was increased and uric acid excretion was decreased, leading to hyperuricemia [12]. Mineo et al. suggested that muscle exercise caused elevated purine metabolite concentrations in the plasma by increasing the flow of purine nucleotides from muscle tissue to plasma, thus leading to hyperuricemia due to increased uric acid synthesis (myogenic hyperuricemia) [6]. Puig et al. presented a patient diagnosed with McArdle's disease while on colchicine and allopurinol treatment for gout disease. The authors attempted to explain the relation between GSDV and hyperuricemia. To this end, they recruited that patient's brother, who had gout with tophus, and six other patients with gout. After administering adenine to the subjects, they measured uric acid, hypoxanthine, xanthine, and creatinine concentrations in plasma and 24-h urine under certain conditions. Moreover, based on the hypothesis that muscle ATP degradation might contribute to the uric acid metabolism, they investigated whether muscle effort would result in a higher increase in the degradation of purine nucleotide in

Table 1 Results of patients with GSDV + gout/hyperuricemia in the literature and the results of our patient

	Our case	Puig et al. [2]	Hardiman et al. [3]	Scalco et al. [7]	Jinnai et al. [5]	Mineo et al. [6]	Hara et al. [8]
Age/sex	35/F	58/M	49/M	50/M	28/M	45/M	45/M 16/M
Diagnosis	GSDV + Gout	GSDV + Gout/renal stone	GSDV + Gout	GSDV + Gout	GSDV + Hyperuricemia	GSDV + Hyperuricemia	GSDV + Hyperuricemia/ GSDV + Hyperuricemia
Symptom on initial admission	Oligoarthritis	Elevated CK and gouty arthritis (23 years of gout history)	Muscle pain, swollen muscles	Exercise intolerance	Muscle stiffness, cramps	Muscle pain, difficulty in physical exercise	Exercise intolerance/ Exercise intolerance
Age at diagnosis of GSDV, years	34	58	49	50	28	Unspecified	Unspecified
The diagnosis/age at diagnosis prior to GSDV, years	Undifferentiated arthritis/25	Gout/35	–	Gout/16	–	–	Unspecified
Condition leading to diagnosis of myopathy	Elevated CK while on colchicine treatment	Elevated CK while on colchicine treatment	Muscle pain, swollen muscles, elevated CK	Rhabdomyolysis attacks and elevated CK	Muscle cramps and stiffness	Muscle pain, difficulty in physical exercise	Unspecified
Second wind phenomenon	+	Unspecified	Unspecified	+	None	Unspecified	–
Family history	None	1 brother with gout with tophus	Unspecified	Unspecified	None	Unspecified	–
Consanguineous marriage between parents	+	Unspecified	Unspecified	Unspecified	None	Unspecified	–
Organomegaly	None	Unspecified	Unspecified	Unspecified	None	Unspecified	–
Delay in diagnosis	10 years	23 years	6 years	About 24 years	Approximately 10–15 years	Unspecified	–
Detection of hyperuricemia (Before or after GSD etc.)	1 year prior to the diagnosis of GSD	23 year prior to the diagnosis of GSD	Simultaneous	24 years before GSD diagnosis	Simultaneous	Unspecified	45 years/16 years
Age at gout diagnosis/-hyperuricemia detection	33 years/–	33 years/–	–/49 years	–/16	–/28 years	Unspecified	–/45 years –/16 years
Time from diagnosis of gout/hyperuricemia to GSDV	1 years	23 years	Simultaneous	24 years	Simultaneous	Unspecified	Unspecified
EMG at the time of GSDV diagnosis	Normal	Myopathy	Unspecified	Unspecified	Unspecified	Unspecified	Unspecified
Muscle biopsy	Glycogen build-up Phosphorylase enzyme negative	Muscle phosphorylase negative	Deficiency of phosphorylase activity in biopsy	Accumulation of glycogen Lack of muscle phosphorylase activity	Intermyofibrillar glycogen granules as shown by electron microscopy	Unspecified	Muscle biopsy positive / Muscle biopsy positive
Enzyme level		Unspecified		Unspecified		Unspecified	

Table 1 (continued)

	Our case	Puig et al. [2]	Hardiman et al. [3]	Scalco et al. [7]	Jinnai et al. [5]	Mineo et al. [6]	Hara et al. [8]
	Myophosphorylase enzyme deficiency		Myophosphorylase deficiency		Phosphorylase alpha deficiency		Lack of enzyme (+)/Lack of enzyme (+)
Mutation	p. Gly686Arg (homozygous) (c.2056G > A) mutation (PYGM)	Unspecified	Unspecified	PYGM (+) (Homozygous R50X)	Unspecified	Unspecified	Unspecified
Forearm ischemic test	Not done	Unspecified	Positive	Positive	Positive	Positive	Positive/Positive
At the time of GSDV diagnosis							
CK level	1200 IU/mL (*N* < 190)	40.01 mkat/L (max 95) ((N:2.16)	211 IU/L (*N* < 70)	1223 IU/L	837 IU/mL	Unspecified	Unspecified
ESR mm/h/CRP mg/dL	40/0.344	–/–	–/–	–/–	–/–	–/–	
Uric Acid evel	5.5 (while on treatment)	8 mg/dL	513 mmol/L (normal>420)	Unspecified	7.7–9.9 mg/dL	7.2–12.7 mg/dL	
Renal excretion of uric acid	–	524 mg/day	Unspecified	Unspecified	5.7–10.5% (normal)	537 mg/day	Unspecified
GSDV treatment	Dietary recommenda-tions	Unspecified	Unspecified	Unspecified	Unspecified	Unspecified	Unspecified
Hyperuricemia/gout treatment	Febuxostat Anakinra	Allopurinol Colchicine	Unspecified	Unspecified	Unspecified	Unspecified	Unspecified
Comment (on the association of GSDV and gout/hyperuricemia)	Myogenic hyperuricemia	Patient has an impaired renal excretion of uric acid Co-existence of gout and GSDV is coincidental	Hyperuricemia is due to increased urate production rather than interference with excretion. (Myogenic hyperuricemia)	Unspecified	Hyperuricemia due to exercise-induced production (Myogenic hyperuricemia)	Myogenic hyperuricemia (exercise-induced overproduction)	Myogenic hyperuricemia (exercise-induced overproduction)

ESR erythrocyte sedimentation rate. *CRP* C-reactive protein, *CK* creatine phosphokinase, *GSDV* glycogen storage disease type V, *EMG* electroneuromyography, *M* male, *F* female

a patient with McArdle's disease compared with that patient's brother with primary gout disease. In all subjects, they found comparable urinary uric acid excretion and plasma purine levels. Plasma hypoxanthine and xanthine concentrations reached higher levels in the patient with McArdle's disease than in the brother with primary gout. Such dramatic modifications in uric acid precursors did not affect serum uric acid concentrations, which remained stable in both siblings during ischemic exercise [2]. Jinnai et al. showed that purine metabolites increased transiently upon exercising and the increment in uric acid persisted until the next day [5].

It has been reported that McArdle's disease may be associated with some immunologic abnormalities. This may lead to misdiagnosis and inappropriate treatment [15]. None of the five age- and sex-matched patients with polymyositis, with whom our one index case was compared, had elevated uric acid values (Table 2). In patients with GSD affecting the skeletal muscle, conversion of adenosine diphosphate (ADP) to ATP decreases due to disrupted glycolysis. In this way, ADP accumulates and triggers hyperuricemia via a purine decomposition pathway [6]. In light of these findings, including the constantly high uric acid concentrations of our patient, we believe hyperuricemia (gout) that arises during the course of GSDV is myogenic hyperuricemia rather than being just coincidental. Elevated muscle enzymes detected during colchicine use in the rheumatic setting are mostly attributed to the myotoxic characteristics of colchicine. This may cause elevated enzyme concentrations to be misinterpreted and any underlying muscle disease to be overlooked, but equally, they may facilitate diagnosis by making sub-clinical metabolic myopathy become overt. Therefore, attention must be paid to elevated muscle enzyme levels in patients while on colchicine.

In conclusion, although findings and symptoms such as hypoglycemia, hypertriglyceridemia, lactic acidosis, and hepatomegaly more easily suggest the likelihood of an underlying metabolic disorder, in the absence of above-listed symptoms, particularly in young (premenopausal female) gout patients, the presence of hyperuricemia alone must be a clue for physicians dealing with musculoskeletal problems to predict an underlying metabolic disease such as GSD. In the presence of hyperuricemia/gout and myopathy, myogenic hyperuricemia must be kept in mind and the patient must undergo the required biochemical and histopathologic analyses for the diagnosis of GSD.

Table 2 Clinical and laboratory findings of GSDV + gout patient and our age- and sex-matched patients with polymyositis

	Our patient	Control patients (polymyositis) (mean ± SD)
N	1	5
Age, years	35	33 ± 9.02
Age at myopathy diagnosis, years	34 (age at onset symptom: 25)	28 ± 4.9
Delay in diagnosis	10 years	3.2 ± 1.64 months
Sex	F	100% female
Laboratory findings during diagnosis of myopathy		
Hb/mm^3	12.9	11.9 ± 1.25
WBC/mm^3	6900	6180 ± 1070
PLT/mm^3	254,000	330,200 ± 105,207
ESR mm/h	40	53 ± 46.6
CRP mg/dL	0.344	10.5 ± 13.5
CK IU/L	2132 (highest value)	5003 ± 6492 (1261–16,443)
LDH IU/L	253	1171 ± 434 (592–1518)
Uric acid mg/dL	13.1 (highest value)	4.42 ± 0.38 (3.90–4.90)
ANA positivity, *n*, %	–	3, 60%
EMG (myopathic pattern) positivity, *n*, %	Normal	3, 60%
Muscle biopsy findings	Glycogen accumulation in PAS Enzyme histochemical and immunohistochemical analysis for phosphorylase was negative	Endomysial mononuclear inflammation and myonecrosis in all patients MHC-I/CD8 positivity in 3 patients PAS negativity in all patients

Hb hemoglobin, *WBC* white blood cell, *ESR* erythrocyte sedimentation rate, *CRP* C-reactive protein, *CK* creatine phosphokinase, *LDH* lactate dehydrogenase, *GSDV* glycogen storage disease, *PM* polymyositis, *EMG* electroneuromyography, *PAS* periodic acid-Schiff staining, *ANA* antinuclear antibodies

Authors' contributions Döndü ÜSKÜDAR CANSU: Contributed to project design, writing, editing, and critical revision of the manuscript and has read and approved the manuscript.

Bahattin ERDOĞAN: Contributed by drafting the manuscript, has read and approved the manuscript.

Cengiz KORKMAZ: Contributed to project design, editing, and critical revision of the manuscript and has read and approved the manuscript.

Compliance with ethical standards

Disclosures None.

Ethical approval All procedures performed in studies involving human participants were in accordance with the ethical standards of the institutional and/or national research committee and with the 1964 Declaration of Helsinki and its later amendments or comparable ethical standards.

Informed consent Informed consent was obtained from our patient included in the study.

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