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【 CASE REPORT 】

Acute Coronary Syndrome Developed in a 17-year-old Boy with Sitosterolemia Comorbid with Takayasu Arteritis: A Rare Case Report and Review of the Literature

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Abstract:

A 17-year-old boy with acute coronary syndrome was admitted to our hospital. He had xanthomas over his elbow and Achilles tendon and a high level of low-density lipoprotein cholesterol; therefore, his initial diagnosis was familial hypercholesterolemia. However, a genetic analysis revealed a compound heterozygous mutation in the *ABCG5* gene with a high serum level of sitosterol, leading to the diagnosis of sitosterolemia. After lipid-lowering treatment, percutaneous coronary intervention was performed. Furthermore, a persistently high C-reactive protein level and images of large arteries led to a diagnosis of Takayasu arteritis. To our knowledge, this is the first case of sitosterolemia complicated by Takayasu arteritis.

Key words: *ABCG5*, heterozygous mutation, low-density lipoprotein cholesterol, C-reactive protein

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Introduction

Sitosterolemia is a rare autosomal recessive sterol storage disorder included in hereditary dyslipidemias that results from mutations in the genes encoding adenosine triphosphate-binding cassette transporters, namely *ABCG5* and *ABCG8* (1). It is characterized by increased plant sterol levels, xanthomas, and accelerated atherosclerosis. Hematologic manifestations including hemolytic anemia with stomatocytosis, macrothrombocytopenia, splenomegaly, and abnormal bleeding can also be associated with it (2). The incidence of heterozygous loss-of-function mutations in the *ABCG5* and *ABCG8* genes has been recently reported to be as high as 1 in 220 (3).

Takayasu arteritis (TA), which is also known as "pulseless disease" or "aortitis syndrome," is a rare disease. A recent systematic review reported an incidence rate of 1.11 cases per million-person years (95% confidence interval: 0.70-1.76) (4). TA is a chronic idiopathic inflammatory disease that predominantly affects large vessels, the aorta, and its major branches, including the coronary, carotid, pulmonary, and renal arteries. Vascular inflammation leads to intimal proliferation of the arterial wall, resulting in lumen stenosis and occlusion. Conversely, inflammation involving the elastic lamina of the arteries may result in an aneurysm (5, 6). The most common manifestations of TA in the coronary arteries include stenosis or occlusion of the coronary ostia, diffuse or focal coronary arteritis, and coronary artery aneurysm, all of which can cause unexpected acute coronary

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Table 1. Laboratory Data of the Patient on Admission.

Blood count		**Biochemistry**		**Lipid profile**	
Red blood cell	461×10⁴ /μL	AST	15 U/L	Total cholesterol	440 mg/dL
Hemoglobin	12.3 g/dL	ALT	19 U/L	LDL-cholesterol	379 mg/dL
Hematocrit	37.5 %	γ-GTP	33 U/L	HDL-cholesterol	35 mg/dL
White blood cell	5,900 /μL	LDH	186 U/L	Triglyceride	108 mg/dL
Platelet	13.5×10⁴ /μL	Alkaline phosphatase	363 U/L	Blood sugar	92 mg/dL
MPV	13.6 fl	Total Bilirubin	0.6 mg/dL	HbA1c	5.5 %
PDW	22.2 fl	Creatin kinase	95 U/L	Apo A-I	98 mg/dL
IPF	5.8 %	Total protein	7.6 g/dL	Apo B	219 mg/dL
Coagulation		Albumin	4.1 g/dL	RLP-C	14.3 mg/dL
APTT	33.0 s	Creatinine	0.71 mg/dL	Lipoprotein (a)	37.6 mg/dL
PT-INR	1.14	BUN	15 mg/dL	α-linolenic acid	63.2 μg/mL
D-dimer	0.8 μg/mL	Na	137 mmol/L	Arachidonic acid	255.6 μg/mL
Serology		K	4.1 mmol/L	Eicosapentaenoic acid	29.9 μg/mL
hs-Toroponin T	0.235 ng/mL	Cl	101 mmol/L	Docosahexaenoic acid	127.5 μg/mL
hs-C-reactive protein	3.493 mg/dL	Uric acid	6.6 mg/dL		
NT-proBNP	385.4 pg/mL				

MPV: mean platelet volume, PDW: platelet distribution width, IPF: immature platelet fraction, APTT: activated partial thromboplastin time, PT-INR: prothrombin time-international normalized ratio, hs: high-sensitive, NT-proBNP: N-terminal fragment of pro-B-type natriuretic peptide, AST: aspartate aminotransferase, ALT: alanine aminotransferase, γ-GTP: γ-glutamyltransferase, LDH: lactate dehydrogenase, BUN: blood urea nitrogen, Na: sodium, K: potassium, Cl: chloride, LDL: low-density lipoprotein, HDL: high-density lipoprotein, HbA1c: hemoglobin A1c, Apo A-I: apolipoprotein A-I, Apo B: apolipoprotein B, RLP-C: remnant like particles-cholesterol

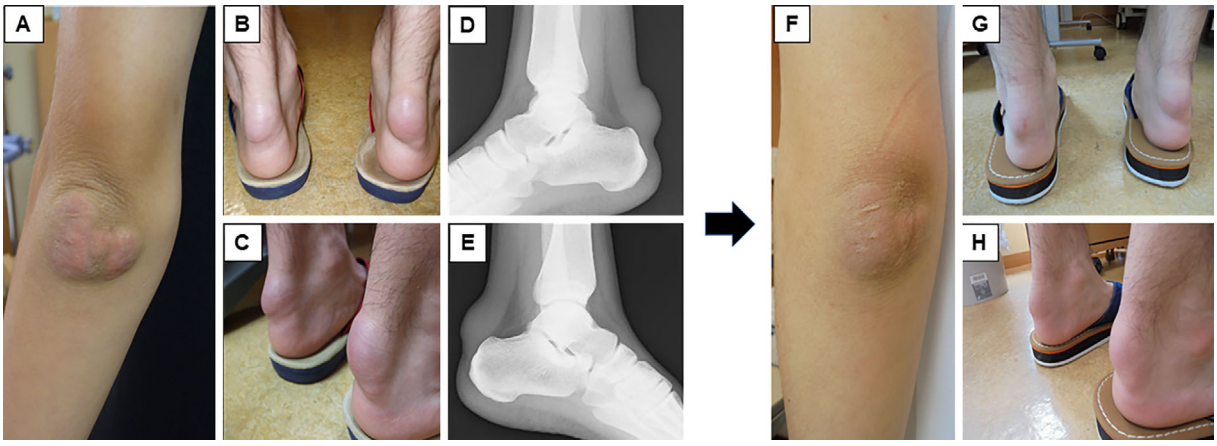

Figure 1. Xanthomas over the elbows and Achilles tendons before and three months after treatment and a radiograph of the ankle. Xanthomas over the elbows before (A) and after (F) treatment. Xanthomas over the Achilles tendons before (B, C) and after treatment (G, H). Radiography of the ankle shows Achilles tendon thickening (D, right side; E, left side).

syndrome (ACS) in younger populations (7).

We herein report a teenage boy who was diagnosed with sitosterolemia caused by a compound heterozygous mutation in *ABCG5* and TA after clinically presenting with ACS.

Case Report

A healthy 17-year-old high school boy with acute-onset chest pain was referred to our hospital. He had no significant medical or family history, and his parents showed no evidence of consanguineous marriage.

Electrocardiography revealed a negative T wave in leads II, III, and aVF and ST elevation in lead aVL. Biochemical tests revealed high levels of cardiac troponin T (0.235 ng/mL), high-sensitivity C-reactive protein (CRP, 3.493 mg/dL), and low-density lipoprotein cholesterol (LDL-C; 414 mg/dL; Table 1). On a physical examination, xanthomas were found over the elbows and Achilles tendons (Fig. 1A-C). A systolic murmur of Levine IV was heard at the apex, and the bruit was audible bilaterally in the carotid and subclavian arteries. Radiography of the ankle showed thickening of the Achilles tendon (right side, 20 mm; left side, 19 mm; Fig. 1D, E). Echocardiography revealed moderate mitral valve regurgitation due to mitral valve prolapse at the A2 and A3 sites and no obvious left ventricular asynergy. Coronary angiography (CAG) revealed 99% stenosis in segment

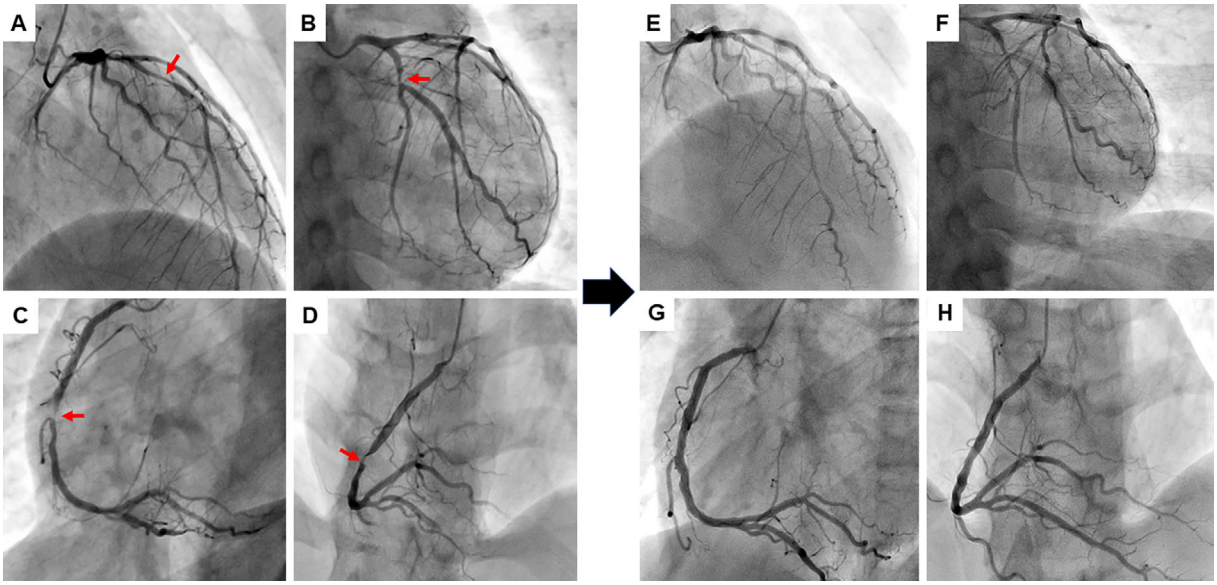

Figure 2. Coronary angiography. The left panel shows 75% stenosis in the left descending artery (LAD) (A), 75% stenosis in the left circumflex artery (LCX) (B), and 99% stenosis in the right coronary artery (RCA) (C, D) on initial coronary angiography. Arrows indicate the stenosis sites. The right panel shows slight regressions in LAD (E) and LCX (F) and the absence of restenosis in the RCA (G, H).

2 of the right coronary artery (RCA) and 75% stenosis in segments 7 and 11 of the left anterior descending (LAD) and left circumflex coronary arteries (Fig. 2A-D). An initial diagnosis of ACS with familial hypercholesterolemia (FH) was made because the patient met the diagnostic criteria of FH established by the Japan Atherosclerosis Society (8).

Considering the patient's age, lifestyle, and high levels of LDL-C, percutaneous coronary intervention (PCI) was planned for the RCA instead of coronary artery bypass grafting after administering lipid-lowering treatment. First, rosuvastatin was initiated at a daily dose of 5 mg and titrated to a maximum daily dose of 20 mg. Subsequently, ezetimibe (10 mg daily), probucol (1,000 mg daily), and evolocumab (420 mg subcutaneous injection every 2 weeks) were sequentially administered for 1 month to achieve a rapid reduction in LDL-C levels; an LDL-C level of <180 mg/dL was achieved on day 31. PCI of the RCA was performed on day 31 based on the findings of myocardial scintigraphy, which indicated inferior wall ischemia. Optical coherence tomography (OCT) revealed the accumulation of macrophages, healed plaque (layered pattern), cavity of the ruptured plaque, and large lipid plaques at the culprit lesion and at the proximal end of the RCA (Fig. 3). During PCI, the culprit lesion was first dilated using a scoring balloon, followed by a drug-coated balloon.

After PCI, the patient's chest pain completely resolved, but high CRP levels persisted. He occasionally had a high fever, and his general condition was poor. Screening for bacterial infection using blood and urine cultures and connective tissue diseases revealed negative findings. Contrast-enhanced computed tomography demonstrated global wall thickening in the aortic arch and its three branches (Fig. 4A), while magnetic resonance imaging revealed the presence of a plaque extending from both the common carotid arteries to the internal carotid artery (Fig. 4B), which led to the diagnosis of TA according to the JCS 2017 Guideline on Management of Vasculitis Syndrome (9). On day 42, the patient was discharged with an LDL-C level of 58 mg/dL. At 3 months after discharge, his LDL-C level decreased to 38 mg/dL, and his xanthomas regressed (Fig. 1F-H).

The patient was readmitted for the treatment of TA six months after his first admission. Prednisolone was initiated at a dose of 50 mg/day. After the addition of tocilizumab (TCZ), his CRP level normalized. Subsequently, fluorodeoxyglucose positron emission tomography demonstrated faint accumulation over the vascular wall of the aortic arch, bilateral common carotid arteries, subclavian arteries, and brachial arteries; however, pretreatment data were unavailable (Fig. 4C). Prednisolone was maintained at a dose of 5 mg/day. CAG performed at 18 months revealed slight regression of the stenosis in the coronary arteries (Figs. 2E-H). However, intracoronary imaging was not performed as the initial OCT findings showed a vulnerable plaque, and there was no progression of stenosis on CAG. The levels of LDL-C and CRP continued to remain low for over 3 years after the onset of ACS (<30 mg/dL and <0.01 mg/dL, respectively), and there was no recurrence of clinical symptoms.

During the second hospitalization, a decrease in the platelet count to 70,000-100,000/μL was observed with the presence of macrothrombocytes (Fig. 5). In addition, intestinal bleeding occurred, although the bleeding site could not be identified by upper or lower gastrointestinal endoscopy, so

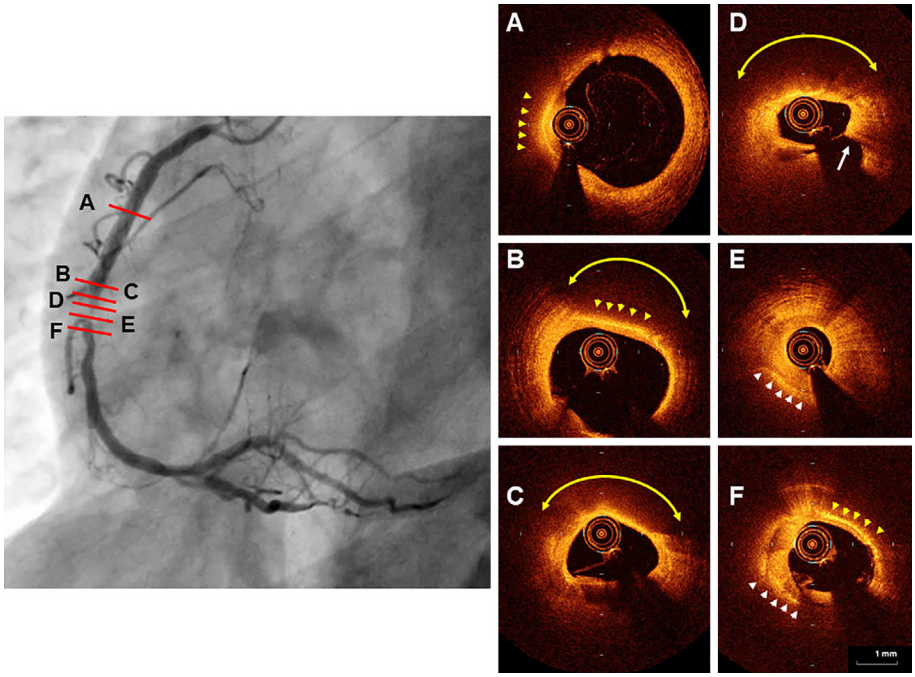

Figure 3. **Findings of optical coherence tomography (OCT) in the right coronary artery. Left panel: The sites (A-F) assessed with OCT in the right coronary artery on coronary angiography. Right panel: OCT images at each site. Yellow arrowheads denote the accumulation of macrophages (high density), white arrowheads denote healed plaque (layered pattern), the white arrow denotes the cavity of the ruptured plaque, and the arced arrow denotes lipid plaque (low density).**

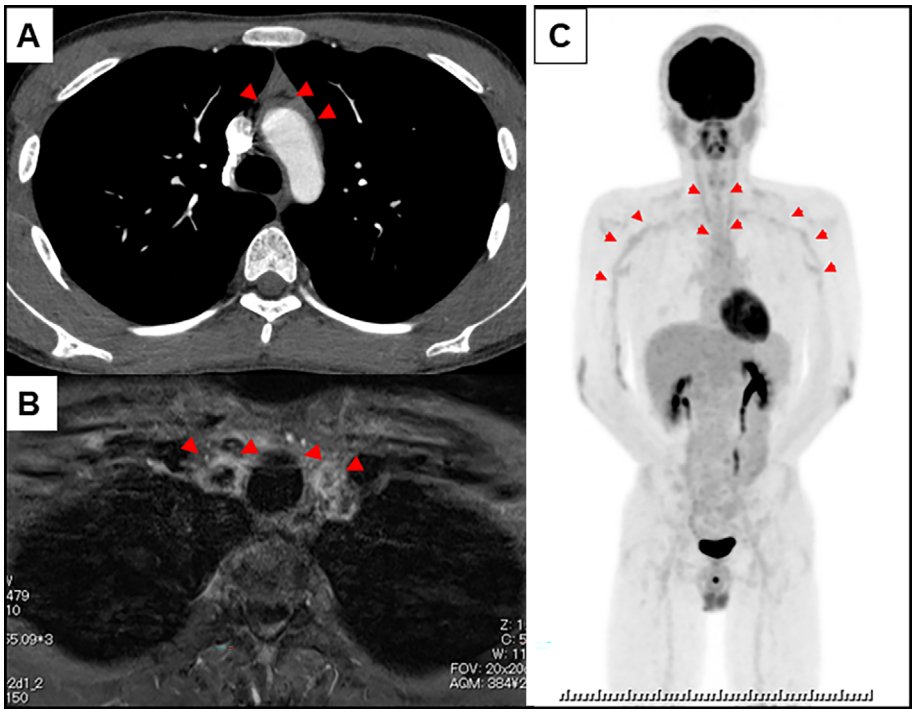

Figure 4. **Images that illustrate the findings of Takayasu arteritis. Wall thickening is found in the aortic arch on contrast-enhanced computed tomography (A) and in carotid arteries on magnetic resonance imaging (B). Fluorodeoxyglucose positron emission tomography (FDG-PET) revealing the faint accumulation in the vascular wall of the aortic arch and bilateral common carotid arteries, subclavian arteries, and brachial arteries (C). Arrowheads indicate wall thickening (A, B) and accumulation on FDG-PET (C).**

clopidogrel was discontinued. Fig. 6 shows the patient's clinical course, including changes in the LDL-C and CRP levels and medical treatment.

Genetic and clinical diagnoses of sitosterolemia

We performed a genetic analysis due to the extremely high level of LDL-C, although the patient's parents had no history of dyslipidemia. Consent from the patient and approval from the Nagasaki University Hospital Ethics Committee (approval number: 20061503) were sought. A total of 21 dyslipidemia-related Mendelian genes, including 3 FH genes (*LDLR*, *PCSK9*, and *APOB*) and other LDL-altering genes (*ABCG5*, *ABCG8*, *APOE*, and *LDLRAP1*), were sequenced. We identified two genetic mutations, a missense mutation (c.1256 G>A or p.Arg419His), and a splice mutation (c.904+1 G>A) in *ABCG5*. Concurrently, the measurement of plasma plant sterol levels demonstrated high concentrations of sitosterol (40 μg/mL; reference range, 1.67-3.13 μg/mL) and campesterol (25 μg/mL; reference range, 2.65-4.45 μg/mL). Based on these findings, the patient was diagnosed with sitosterolemia.

At this point, the patient's family has not consented to a genetic analysis of any further members.

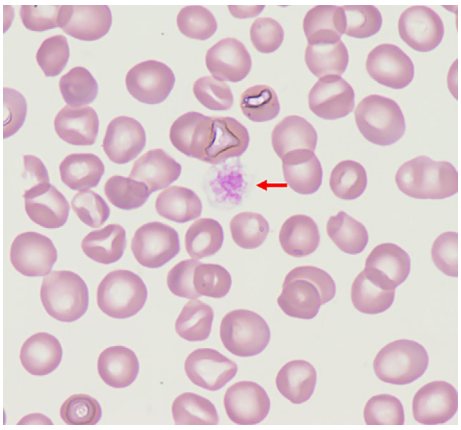

Figure 5. Smear of peripheral blood. The red arrow indicates a macrothrombocyte, which is larger than erythrocytes.

Discussion

To our knowledge, this is the first reported case in which sitosterolemia complicated by TA led to the development of ACS. In addition, the splice mutation of *ABCG5* was a novel finding. Given that lipids and inflammation are considered key drivers of atherosclerotic plaque growth and rupture, our patient with extremely high levels of LDL-C and CRP was at a very high risk of CAD.

We initially misdiagnosed the patient with FH due to an extremely high LDL-C level and the concomitant presence of xanthomas and premature CAD. Patients with sitosterolemia have heterogeneous clinical phenotypes that range from asymptomatic to severe hypercholesterolemia with accelerated atherosclerosis and premature CAD. Varying serum LDL-C concentrations ranging from normal to extremely high have been reported in patients with sitosterolemia, although previous reports have shown that these patients do not usually exhibit a profound elevation in LDL-C levels (10-12). Highly elevated LDL-C levels are often observed in patients with infantile disease. We previously encountered 4 patients with infantile sitosterolemia with 2 pairs of mutations in the *ABCG5* gene who had high levels of LDL-C, ranging from 407 to 796 mg/dL (13). In a case reported by Ono et al., an 18-month-old patient with compound heterozygous mutations in the *ABCG5* gene had extremely high levels of LDL-C (679 mg/dL) and sitosterol

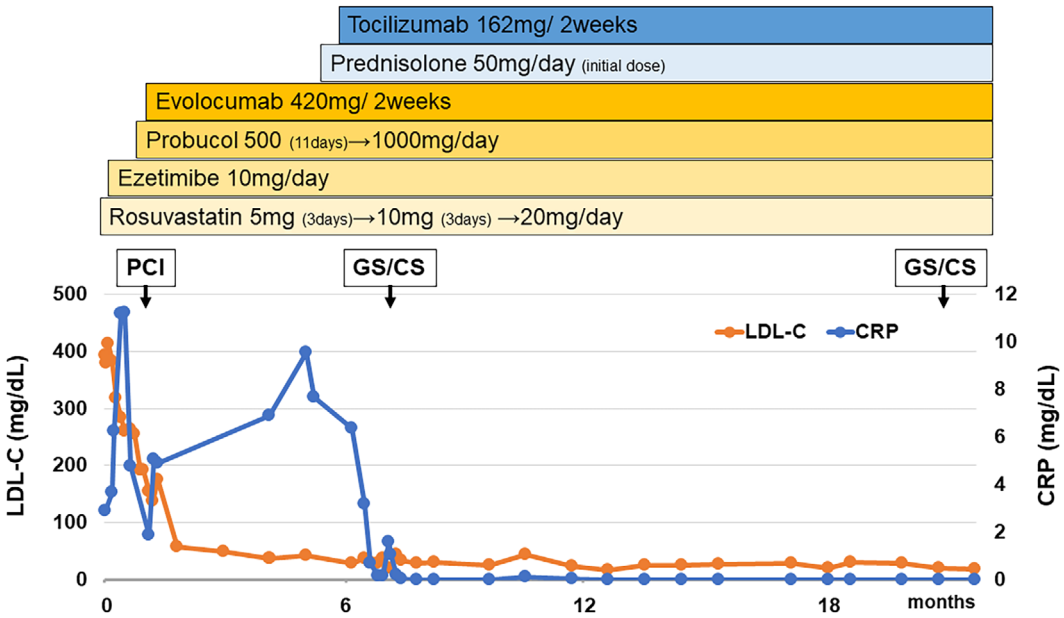

Figure 6. Clinical course of the patient. PCI: percutaneous coronary intervention, GS/CS: gastroscopy/colonoscopy

(24.6 mg/dL) (14). However, in another case report, the median LDL-C level among four patients with sitosterolemia (average age: 18 years) with double mutations in the *ABCG5* or *ABCG8* gene was 154 mg/dL (15). Mymin et al. reported that the cholesterol levels ranged from 139 to 411 mg/dL in 21 patients (age: 1.7-54 years old) homozygous for the *ABCG8 S107X* mutation (16). Table 2 shows the levels of LDL-C and sitosterol in previous cases with mutations in the *ABCG5* gene. Based on these cases, we concluded that the LDL-C and sitosterol levels and clinical features did not differ markedly between patients with compound heterozygous and homozygous *ABCG5* mutations.

The modulation of cholesterol metabolism in patients with sitosterolemia has not yet been elucidated. In a large cohort of 207,926 subjects, LDL-C concentrations were significantly associated with absolute concentrations of non-cholesterol sterols, such as lathosterol, desmosterol, β-sitosterol, campesterol, and cholestanol. In addition, the relative concentrations of the cholesterol synthesis markers lathosterol and desmosterol were positively correlated with the concentration of LDL-C, whereas those of the cholesterol absorption markers β-sitosterol, campesterol, and cholestanol were inversely correlated with the concentration of LDL-C, suggesting that both cholesterol synthesis and absorption may regulate LDL-C concentrations (17). Furthermore, an increase in plant sterols in hepatocytes inactivates sterol regulatory element-binding protein 2. Consequently, the hepatic expression of LDL-receptor is downregulated, which leads to a decrease in the uptake of serum LDL-C and an elevation in cholesterol levels (18). TA occurred as a complication in our patient. Wang et al. demonstrated that patients with active TA had proatherogenic lipid profiles, as represented by reduced levels of apolipoprotein A1 (apoA1) and high-density lipoprotein cholesterol as well as increased ratios of apolipoprotein B (apoB) to apoA1, compared to 132 premenopausal female patients with TA and 100 healthy controls matched for sex, age, and body mass index (19). This may explain the high levels of LDL-C in our patient.

Regarding inflammation, CRP has been reported to be a predictor for the risk of adverse cardiovascular events and to be involved in atherogenesis (20, 21). In addition, several recent large cardiovascular outcome trials have demonstrated that anti-inflammatory drugs, such as canakinumab (a monoclonal antibody of IL-1β) and colchicine, reduce the risk of cardiovascular events (22-24). Thus, TA-derived inflammation and the high level of LDL-C may have been responsible for the advanced atherosclerotic plaque formation in our patient. However, how sitosterol, LDL-C, CRP and other factors contribute to this formation is unclear.

The development of premature CAD has been reported in patients with sitosterolemia and in those with TA. Coronary stenosis in patients with TA is often located at the ostium or in the proximal segments of the coronary artery (7). However, the characteristics of the coronary lesions associated with sitosterolemia have not been fully elucidated. We reviewed previous case reports that included images obtained during CAG or descriptions of coronary lesions. We determined the site and rate of stenosis using the CAG images, if these descriptions were not available in the reports (Table 3). Coronary stenotic lesions in patients with sitosterolemia can be located in the proximal or mid segments, and multi-vessel lesions are more common than single-vessel lesions. In our case, the coronary lesions were located in the proximal and mid portions of the three vessels, suggesting that the lesions had been caused by sitosterolemia. We also evaluated the characteristics of plaques in the coronary arteries using OCT. The findings in our patient were characteristic of atheromatous plaques and included a large lipid-rich plaque, macrophage accumulation, and healed plaque, not only at the culprit lesion but also at distant sites. These findings indicated that atherosclerosis in the coronary arteries was accelerated. Few reports have described the components of plaque in patients with sitosterolemia. Further studies are required to characterize the components of plaques in these patients.

The treatment of patients with sitosterolemia caused by mutations in the *ABCG5* gene is shown in Table 2. In infantile patients, weaning from breastfeeding is an effective treatment option, as breast milk contains around 100 mg/dL of sitosterol. If an infant ingests 1,000 mL of breast milk per day, then the infantile consumption of sitosterol is estimated to be around 1,000 mg/day; this amount is close to that of an adult and therefore is too much for an infant to safely consume (13, 25). LDL-C-lowering drugs have been used to treat patients with sitosterolemia. Among these drugs, ezetimibe and colestimide are particularly effective in reducing cholesterol and sitosterol levels (26). However, whether or not the normalization of the concentration of plant sterols is essential for preventing CAD remains unclear. A meta-analysis found no significant association between sitosterol levels and the risk of CAD (27). The risk of CAD in heterozygous carriers of the loss-of-function mutation in the *ABCG5* gene was found to be proportional to the effect of LDL-C elevation (28). Therefore, LDL-C levels may be a key regulator of atherosclerosis in patients with sitosterolemia. We sequentially administered LDL-C-lowering drugs, such as statins, ezetimibe, probucol, and PCSK9 inhibitors, for six weeks because of the initial diagnosis of FH in our patient. The combination therapy resulted in an LDL-C level of approximately 30 mg/dL. This aggressive treatment may have caused the relatively rapid regression of xanthomas in our patient. Previous reports showed that such regression was observed 9 months to 1.5 years after treatment, although the patients had different backgrounds and treatment strategies (29-32).

The effect of PCSK9 inhibitors on sitosterol levels has not yet been established. A few studies have reported the effectiveness of PCSK9 inhibitors in patients with sitosterolemia, especially those with high levels of cholesterol (33, 34). Thus, statins and PCSK9 inhibitors can be considered treatment options for patients with very high levels of LDL-C, although statins are not deemed effective in

Table 2. LDL-C and Sitosterol Levels, Clinical Features, and Treatment in Patients With *ABCG5* gene Mutation.

Ref.	Age	Sex	LDL-C (mg/dL)	Sitosterol (μg/mL)	*ABCG5* gene mutation	Clinical features	Treatment
Heterozygous mutation							
Our case	17y	M	414	40.0	c.1256G>A; p.R419H c.904+1G>A	ACS, xanthoma, macrothrombocytopenia, gastrointestinal bleeding	Rosuvastatin, ezetimibe, probucol, evolocumab
37	3m	F	304	91.7	c.1166G>A; p.R389H c.1336C>T; p.R446X	NA	Cholestyramine, ezetimibe
13	10m	F	555 (TC)	33	c.1813_1817delCTTTT; p.P558EfsX14 c.1306G>A; p.R389H	NA	Weaning from breastfeeding, ezetimibe
38	11m	F	837	23.7	c.47C>T; p.Q16X c.1336C>T; p.R446X	Xanthoma	Statins, ezetimibe→ ezetimibe, rosuvastatin, weaning from breastfeeding
13	13m	F	796	80	c.130T>C; p.S44A c.1306G>A; p.R389H	Xanthoma	Weaning from breastfeeding, diet, ezetimibe
30	15m	F	540	193.6	c.904+1G>A; p.M302Nfs*82 c.1336C>T; p.R446X	Xanthoma	Diet, cholestyramine
14	18m	F	679	246	c.1256G>A; p.R419H c.1166G>A; p.R389H	Xanthoma	Diet, colestimde, ezetimibe
13	18m	F	407	101	c.1813_1817delCTTTT; p.P558EfsX14 c.1306G>A; p.R389H	Xanthoma	Weaning from breastfeeding, diet, ezetimibe
37	18m	F	565	71.0	c.1166G>A; p.R389H c.1336C>T; p.R446X	Xanthoma	Cholestyramine →ezetimibe
39	1y	F	453	15.9	c.1166G>A; p.R389H (*ABCG8* gene c.1285A.C; p.M429L)	Xanthoma	Weaning from breastfeeding, diet
13	24m	M	589	115	c.1813_1817delCTTTT; p.P558EfsX14 c.1336T>C; p.R446X	Xanthoma	Weaning from breastfeeding, colestimide →ezetimibe
40	2y	F	690	78.8	c.751C>T; p.Q251X c.1336C>T; p.R446X	Xanthoma	Diet, ezetimibe
37	8y	F	346	NA	c.987C>A; p.Y329X, c.1311C>G; p.N437K	Xanthoma, hemolytic anemia	Diet, cholestyramine
41	10y	F	224	681	c.144-1G>A; p.H510T c.1523 delC; p.L511X	Xanthoma, Achilles tendon thickness, anemia, macrothrombocytopenia	Diet, cholestyramine, ezetimibe
37	12y	F	263	61.4	c.1166G>A; p.R389H c.850G>A; p.G269R	Xanthoma	Statin →ezetimibe
42	13y	F	609	182	c.1256G>A; p.R419H c.1763-1G>A [splice acceptor site]	Xanthoma, carotid artery plaque, AR with calcification	Rosuvastatin, ezetimibe
43	56y	F	274	3.1*	c.1166G>A; p.R389H (*CD36* gene c.1126-5_1127delTTTAGAT *PCSK9* gene c.2004C>A; p.S668R)	ACS (cardiac arrest, VF)	Rosuvastatin, ezetimibe
Homozygous mutation							
37	23m	F	519	70.7	c.1166G>A; p.R389H	Xanthoma	Ezetimibe
44	5y	F	868 (TC)	94.8	c.1166G>A; p.R389H	Xanthoma, carotid artery plaque	Diet, ezetimibe
45	14y	M	332	214.3	c.1336 C>T; p.R446X	Xanthoma, AS, carotid artery plaque, anemia, thrombocytopenia	Diet, ezetimibe
46	21y	M	152 (TC)	175	c.1336C>T; p.R446X	CAD, WPW syndrome, abdominal-infrarenal aortic plaque, liver cirrhosis, xanthoma, Achilles tendon thickness, hemolytic anemia, macrothrombocytopenia,	Diet, ezetimibe

* Data on 24 hospital day. Diet indicates low cholesterol/plan sterol diet. M: male, F: female, TC: total cholesterol, LDL-C: low density lipoprotein cholesterol, ACS: acute coronary syndrome, AR: aortic valve regurgitation, VF: ventricular fibrillation, AS: aortic valve stenosis, CAD: coronary artery disease, NA: not available

Table 3. The Stenotic Lesion in Coronary Arteries in Patients with Sitosterolemia.

Ref.	Age	Sex	Stenotic lesions in coronary arteries
Our case	17y	M	RCA #2 99%, LAD #7 75%, LCX #11-distal 75%
11	16y	F	LAD OS total, LCX #12, 14 total, RCA #1-mid >70%
47	16y	M	LAD #6-mid 80-90%, LCX #11-mid 70%, RCA #1-mid 50-60%
26	18y	F	RCA #1-2 90%,
48	19y	M	RCA #1-distal total, LAD #6-proximal 99%, #7 90%, LCX #11 90%, #13 90%
46	21y	M	LAD #7 99%, RCA#1 total
49	25y	F	RCA #1-mid total
34	60y	M	LAD #6, HL total, RCA#1 50%

M: male, F: female, RCA: right coronary artery, LAD: left descending artery, LCX: left circumflex artery, HL: high lateral artery, OS: ostium, total: total obstruction

treating sitosterolemia. In our case, TA was well controlled by the combination of prednisolone and TCZ, and coronary artery stenosis showed slight regression after the administration of these drugs. In an 18-year-old woman with TA who had severe stenosis in the ostium of both the left main trunk and the LAD, 4 months of combined immunosuppressive treatment with corticosteroid and TCZ resulted in a regression of the coronary ostial stenosis (35). Pan et al. demonstrated that six-month treatment with tocilizumab improved the degree of lumen stenosis in most lesions of the coronary arteries in patients with TA (36). Thus, the suppression of inflammation by anti-inflammatory drugs for TA may also prevent the progression of atherosclerosis in the coronary arteries.

Conclusion

Our patient with sitosterolemia who had a compound heterozygous mutation in the *ABCG5* gene presented with xanthomas, ACS, hypercholesterolemia, and macrothrombocytopenia, leading to intestinal bleeding. In addition, the patient was diagnosed with TA. Both sitosterolemia and TA are rare diseases; therefore, this case is very rare. Aggressive treatment with lipid-lowering and anti-inflammatory drugs led to regression of coronary atherosclerosis.

The authors state that they have no Conflict of Interest (COI).

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