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# A case of sitosterolaemia with stomatocytanaemia and thrombocytopenia treated with Ezetimibe with good response

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From 7th APPEs Biennial Scientific Meeting  
Nusa Dua, Bali. 14-17 November 2012

Sitosterolaemia is a rare autosomal recessive lipid disorder characterized by increased absorption of plant sterols in the gut and decreased biliary excretion of sterols causing accumulation of plasma sterols, which can lead to premature atherosclerosis.

Here we reported a boy presented with multiple tuberous xanthomas at 4 year old and was diagnosed sitosterolaemia [1]. The fasting plasma total cholesterol and low-density lipoprotein (LDL) cholesterol levels were 18.3 mmol/L and 16.41 mmol/L respectively. Gas chromatography and mass spectrometry showed that the fasting plasma sterols contained elevated beta-sitosterol 880  $\mu\text{mol/L}$  (Reference range  $<12 \mu\text{mol/L}$ ), campesterol 489  $\mu\text{mol/L}$  (Reference range  $<17.5 \mu\text{mol/L}$ ) and stigmasterol 38.9  $\mu\text{mol/L}$  (Reference range  $<3.5 \mu\text{mol/L}$ ). Molecular study identified compound heterozygous mutations (R419H and IVS12+IG $\rightarrow$ A) in the adenosine triphosphate (ATP) binding cassette subfamily G, member 5 (*ABCG5*) gene.

Initial management included dietary restriction in cholesterol and plant sterols and cholestyramine treatment. The total cholesterol and LDL cholesterol levels decreased.

The boy developed bleeding tendency with gum bleeding and epistaxis and hepatosplenomegaly at 7 years old. Blood test confirmed and thrombocytopenia and peripheral smear revealed stomatocytanaemia and giant platelets [2]. Bone marrow study showed hypercellular marrow. He was treated with increasing dose of cholestyramine but the drug compliance was fair. The haematological problems persisted.

At the age of 13, Ezetimibe 10mg daily was added. Ezetimibe blocks the absorption of dietary and biliary sources of cholesterol and plant sterols. The platelet count rose from  $58 \times 10^9/\text{L}$  to  $107 \times 10^9/\text{L}$  in 4 weeks' time and normalized after 10 months of Ezetimibe treatment. The haemoglobin level rose from 11 g/dL to 13.8 g/dL in 4 months. The plant sterol level also showed significant improvement (see table 1). There were decreased liver and spleen size. The drug was well tolerated with no adverse effect. The efficacy of Ezetimibe in our patient was sustained after 4 years of treatment which was consistent with the other studies on long-term Ezetimibe treatment [3-6].

**Table 1**

	2004	2006	Mar 2008	Jul 2008 (Ezetimide started in Jun 2008)	Nov 2008	2009	2010	2011
Haemoglobin (13-17 g/dL)	8.8	8.9	11	10.5	13.8	14.6	14.4	15.7
Platelet ( $150-400 \times 10^9/\text{L}$ )	60	59	58	107	100	167	262	124
Campesterol ( $<17.5 \mu\text{mol/L}$ )	489	329	266	-	-	199	196	192
Stigmasterol ( $<3 \mu\text{mol/L}$ )	38.9	31	19	-	-	22.3	24.8	27.6
Beta-sitosterol ( $<12 \mu\text{mol/L}$ )	880	548	617	-	-	443	345	360

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In conclusion, Ezetimibe treatment was effective in lowering the plasma cholesterol and sterols level in our patient with sitosterolaemia. It is also effective in reversing the stomatolyticaemia and thrombocytopenia.

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Published: 3 October 2013

#### References

1. Cheng WF, Yuen YP, et al: Sitosterolaemia and xanthomatosis in a child. *Hong Kong Med J* 2003, **9**:206-209.
2. Rees DC, Iolascon A, et al: Stomatocytichaemolysis and macrothrombocytopenia (Mediterranean stomatocytosis/macrothrombocytopenia) is the haematological presentation of phytosterolaemia. *British Journal of Haematology* 2005, **130**:297-309.
3. Salen G, von Bergmann K, et al: Ezetimibe effectively reduces plasma plant sterols in patients with sitosterolemia. *Circulation* 2004, **109**:966-971.
4. Salen G, Starc T, et al: Intestinal cholesterol absorption inhibitor Ezetimibe added to cholestyramine for sitosterolemia and xanthomatosis. *Gastroenterology* 2006, **130**:1853-1857.
5. Lutjohann D, von Bergmann K, et al: Long-term efficacy and safety of ezetimibe 10mg in patients with homozygous sitosterolemia: a 2-year, open-label extension study. *Int J ClinPract* 2008, **62**:1499-1510.
6. Niu DM, Chong KW, et al: Clinical observations, molecular genetic analysis, and treatment of sitosterolemia in infants and children. *J Inherit Metab Dis* 2010, **33**:437-443.

doi:10.1186/1687-9856-2013-S1-P174

**Cite this article as:** Hung and Lee: A case of sitosterolaemia with stomatocyticanaemia and thrombocytopenia treated with Ezetimibe with good response. *International Journal of Pediatric Endocrinology* 2013 **2013**(Suppl 1):P174.

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