



Thiamine responsive megaloblastic anemia due to *SLCA19A2* gene mutation: Another cause of neonatal diabetes with successful switch from insulin to thiamine

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Introduction:

Thiamine responsive anemia (TRMA) known as Rogers Syndrome; is an early-onset, autosomal recessive disorder characterized by diabetes mellitus, megaloblastic anemia and sensorineural deafness. Diabetes in this condition is well described in infancy but has only very rarely been reported in association with neonatal diabetes.

Case:

- 3-months old male patient with neonatal diabetes
- Uncontrolled hyperglycemia under insulin treatment
- Parental consanguinity (+)
- He was born 2000 grams at 32nd GW
- He was operated on the second day of life due to duodenal atresia
- During hospitalization in intensive care unit, hyperglycemia and supraventricular tachycardia had been detected
- Insulin, propranolol and amiodarone was started
- The patient was discharged 20 at day of life
- Physical examination: On admission he was 56 cm (3p.), 3,4 kg (<3p); head circumference was 38 cm (3p.) and neuro-motor development was consistent with age
- He had been on NPH insulin 0,18 mg /kg /day-divided into two doses
- He had frequent hyper and hypoglycemias during the day
- Blood glucose levels improved after switching to insulin detemir three times a day
- RFX-6 mutation was considered due to the associated of duodenal atresia, and found to be negative
- The patient also had anemia (Hb:8,4 g/dl, MCV:84, MCHC:35,7, ferritin 69ng/dl, Vitamin B12: 168)

Table: Laboratory findings at presentation and 3rd month of thiamine treatment

	Presentation	At Thiamine Tx 3 rd month
Hba1c (%)	7.2	6
Glukose (mg/dl)	277	89
C-peptide (ng/ml)	0.28	1.6
Hemoglobine (g/dl)	8.4	10.1
MCV (fl)	84	69.9
Diabetes Autoantibodies*	Negative	
SLCA19A2	c.242dup; p.Y81X Frameshift homozigous	

Treatment

- Replacement therapy for vitamin B12 deficiency was given. Thiamine replacement was started intramuscularly at first (2x100 mg/day, five times in total) and then continued as 30 mg/kg/day per oral (divided into two doses) after the detection the mutation at *SLCA19A2* gene
- After thiamin replacement, insulin requirement decreased gradually, and insulin therapy was stopped at the end of the first week. Ketosis and hyperglycemia were not observed on the follow-up. He was discharged with oral thiamine
- At the end of three months of thiamine treatment his HbA1c was 6% without insulin

Conclusions

- ✓ TRMA should be kept in mind in the concurrence of megaloblastic anemia, deafness and diabetes
- ✓ Special caution must be paid in terms of arrhythmias that may accompany to disease
- ✓ Genetic researches in the field of neonatal diabetes increase treatment success and change the direction of treatment