Background:
Methylmalonic acidemia (MMA) is an autosomal recessive disorder of metabolism caused by deficient methylmalonyl-CoA mutase activity or impaired transport and synthesis of its cofactor, cobalamin. We present siblings from consanguineous parents with neonatal heterogeneous revelation of the disease: a cardiomyopathy for one and seizures for the second.

Case 1:
A 13 days old boy (Fig.1) with a history of a sister’s death at neonatal period with heart failure, was transferred from pediatric cardiology department for dilated cardiomyopathy with 25% left ventricular ejection fraction (LVEF) and pulmonary hypertension. Results of laboratory studies demonstrated metabolic acidosis, mild elevation of serum lactic acid. Urine organic acid chromatography showed high concentrations of methylmalonic acid and little concentration of methylcitrate. Homocysteine level was 5.36µmol/l (N=5-15µmol/l). Profile of acylcarnitine in blood and urine organic acids was compatible with methylmalonic acidemia. Whole exom sequencing showed methylmalonic aciduria, cblA complementation type.

Conclusions:
These siblings developed two distinct clinical presentations of MMA: an isolated cardiac manifestation and a stroke. Physicians should be aware about these atypical presentations of the disease which can be fatal if not diagnosed and managed rapidly.