**INTRODUCTION**

1. Most children with carbonic anhydrase VA (CA-VA) deficiency reported to date have presented between day 2 of life and age 20 months with hyperammonemic encephalopathy.
2. Four affected individuals have been reported to date.
3. Almost all affected individuals reported to date have shown normal psychomotor development and no further episodes of metabolic crisis.

**CASE REPORT**

A female at age of 3y 7/12, after unremarkable development, presented with 1-day history of fever, diarrhea, insufficient oral intake and lethargy. During intravenous hydration she developed Kussmaul breathing and hyperglycemia (21.8 mmol/l).

At admission in ICU the child was somnolent and fall into coma on day 9. Initial investigations revealed:
- hyperammonemia (421 µmol/l),
- hyperlactatemia (7.24 µmol/l),
- metabolic acidosis that switched to respiratory alkalosis during treatment with dextrose, carbaglumic acid, sodium phenylbutyrate, L-arginine hydrochloride, L-carnitine. Liver function tests remain normal. Blood and urine metabolic testing indicated suspect data for 3-methylcrotonylglycinuria. Clinical and metabolic findings normalized with the treatment.

She continues to have infrequent episodes of vomiting without hyperammonemia or lactic acidosis.

Child demonstrated good developmental progress and remained clinically stable with L-carnitine, low protein diet and "sick-day management" with high-caloric and lipid-rich formula during illnesses.

**RESULTS**

Exome sequencing of 1489 genes for metabolic and mitochondrial disorders identified two heterozygous pathogenic variants in genes CASA (c.555G>A; p.Lys185=), associated with Carbonic anhydrase SA deficiency and SCO2 (c.418G>A; p.Glu140Lys), associated with cytochrome C oxidase deficiency. Additional DNA analysis of CASA gene revealed heterozygous deletion (exone 1) and heterozygous mutation with uncertain significance c.555G>A (silent).

Gas chromatographic profile of organic acids in urine of patient with Carbonic anhydrase VA deficiency and urine from healthy patient

**CONCLUSIONS**

CAVA deficiency should be considered in the differential diagnosis for hyperammonemia and hyperlactatemia in neonate and young children. There is a small number of reported cases (only four) and limited data on long-term follow up. Genotype-phenotype correlations remain to be determined.