

HYPERINSULINAEMIC HYPOGLYCAEMIA AS MEHMO SYNDROME COMPONENT: A CASE REPORT

Diliara Gubaeva, Maria Melikyan
Endocrinology Research Centre, Moscow, Russian Federation



INTRODUCTION

M mental retardation
E epileptic seizures
H hypogonadism
M microcephaly
O obesity

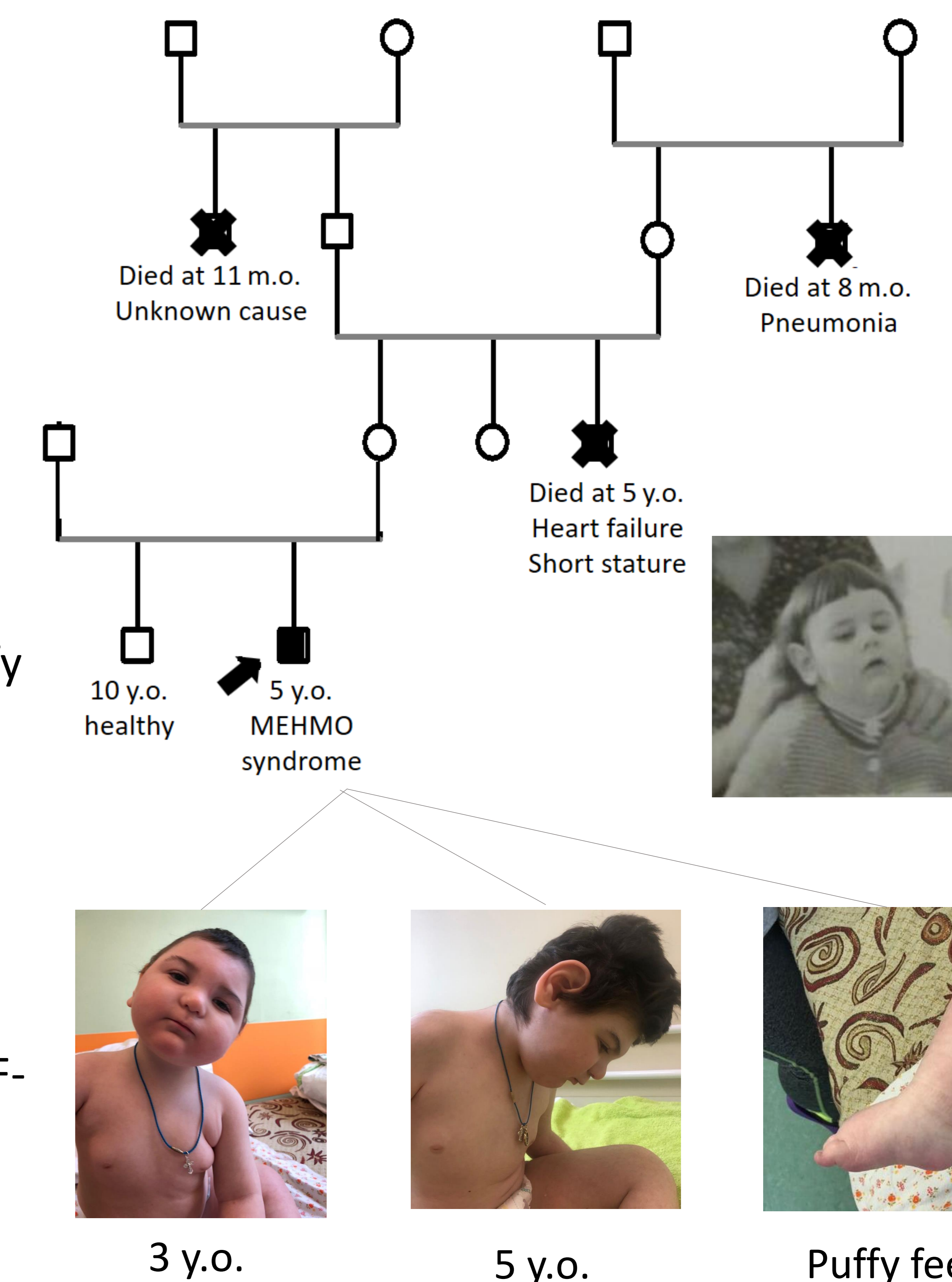
- MEHMO syndrome is a rare X-linked disorder caused by *EIF2S3* gene mutations. This gene encodes a key factor for integrated stress response and initiation of protein synthesis.
- Some of the reported cases of MEHMO syndrome include endocrine disorders: hypopituitarism, diabetes mellitus, and transient hypoglycaemia.
- Here, we present a boy with MEHMO syndrome and hyperinsulinaemic hypoglycaemia.

CLINICAL CASE

- Second child of non-consanguineous Caucasian family.
- At birth: normal weight and length, breathing difficulties.
- 6 months: first seizures, decline in psychomotor development, microcephaly. Diagnosed with epilepsy, started on anticonvulsants.
- 21 months: hyperinsulinaemic hypoglycaemia (glucose 2.2 mmol/l, insulin 28 µU/mL), started on diazoxide 5 mg/kg/day with a good response.
- Severe developmental delay and dysmorphic features: microcephaly, amphora-like face, sloping forehead, macrotia, high-arched palate, dental diastasis, conical teeth, inverted nipples, and puffy hands and feet.
- Brain MRI: cortical-subcortical atrophy, periventricular leukoencephalopathy, thinning of the corpus callosum.
- 3 years: c.671T>G mutation in *EIF2S3* gene (whole genome sequencing).
- 5 years: diazoxide weaned off, no signs of diabetes mellitus or hypopituitarism (cortisol, ACTH, IGF-1, free T4, and TSH levels are within normal range).
- Symptoms associated with MEHMO syndrome in our patient: microcephaly, mental retardation, epileptic seizures, overweight, thin corpus callosum, cryptorchidism, and dysmorphic features.
- Unreported symptoms: 1) hyperinsulinaemic hypoglycaemia; 2) hyperammonemia, alkaline urine, and seizures, improving on low-protein diet.

CONCLUSIONS

- This is the first report of hyperinsulinaemic hypoglycaemia in a patient with MEHMO syndrome.
- Since *EIF2S3* mutations alter protein production, overproduction of insulin is counterintuitive. Presumably, the mechanism resembles MODY1 and MODY3.



CONTACT INFORMATION

gubaevadn@gmail.com

