Congenital hypopituitarism and severe developmental delay associated with homozygous *POU1F1* mutation

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Background

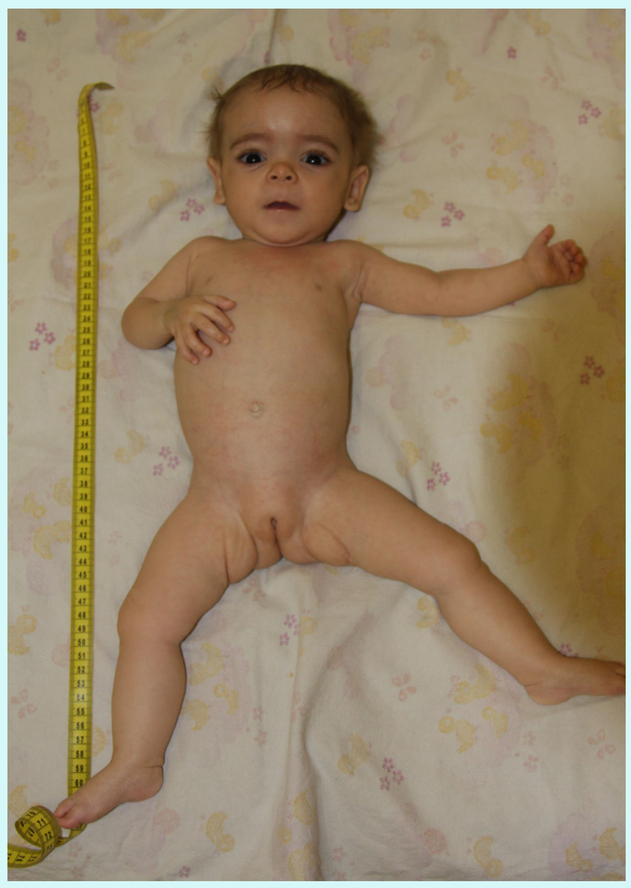
Mutations in *POU1F1* is a rare cause of combined pituitary hormone deficiency, which commonly includes GH, TSH and prolactin deficiencies and characterized by hypoplastic anterior pituitary

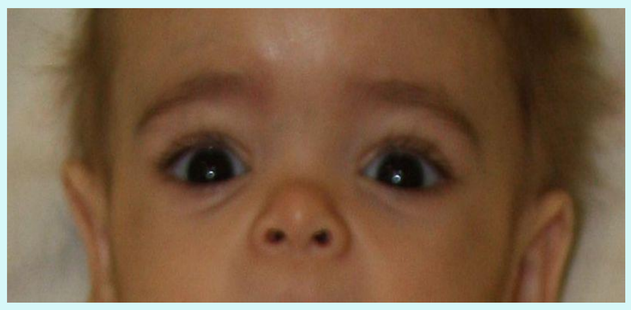
Clinical case:

1.5 year old girl was admitted to our hospital because of short stature

- *Consanguineous healthy parents*
- *Birth length 47 cm, birth weight 2220 g*
- *Psycho-motor delay were noted during first months of life*
- *Had severe developmental delay*
- *Central hypothyroidism was diagnosed at 2 months of age and started on Levothyroxine*

Age 1,5 years
- **Height 58 cm (SDS -7.3)**
- **Weight 3.8 kg (BMI SDS -7.4)**





She had sparse hair, prominent forehead, saddle nose and blue sclera

Laboratory testing revealed:

Asymptomatic ketotic hypoglycemias

IGF-1 3 ng/ml

Prolactin <30 mU/l)

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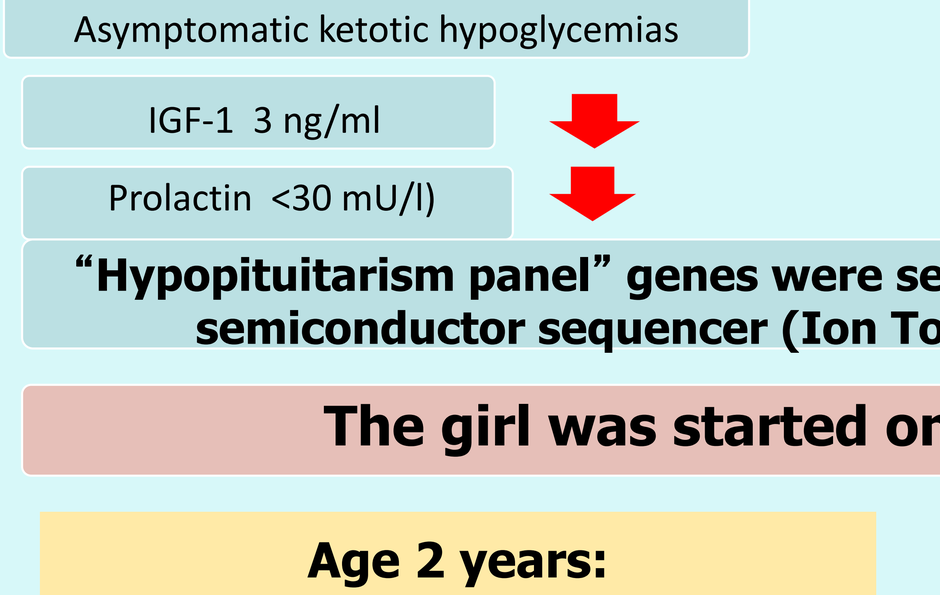
Cortisol 495 nmol/l ACTH 12.7 pg/ml

Brain MRI : anterior pituitary hypoplasia

"Hypopituitarism panel" genes were sequenced using a custom Ion Ampliseq gene panel and PGM semiconductor sequencer (Ion Torrent) Homozygous R256W mutation in *POU1F1* gene

The girl was started on GH therapy and carbohydrate rich diet

Age 2 years:
- **Height 67 cm (SDS -4.98)**
- **Weight 5.5 kg (BMI SDS -5.9)**
- She showed some improvement in psychomotor development



Conclusion:

***POU1F1* mutations is a rare cause of hypopituitarism, which may present with failure to thrive and extremely short stature, showing a good response to GH therapy. Severe developmental delay, seen in our case, may be a result of untreated neonatal hypoglycemia, hypothyroidism or be a part of a syndrome.**

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