

Congenital hypopituitarism in a patient with 18p- syndrome

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OBJECTIVE

Description of a rare clinical case of congenital hypopituitarism as a feature of 18p- syndrome.

BACKGROUND

18p- syndrome is very rare (1:50000 live-born infants) [1]. The main clinical features are short stature, round face with short philtrum, palpebral ptosis, large ears with detached pinnae, intellectual deficiency. Hypopituitarism as part of the syndrome is found in 13% of cases and associated with breakpoint in the centromeric region [2].

RESULTS

A girl to non-consanguineous healthy parents was born at term with normal height and weight. From the first months of life the child had marked muscular hypotonia, dysphagia, recurrent hypoglycaemia and cholestasis (jaundice, hepatomegaly, acholic stool, and biochemical markers (Tab.1, Pic.1).

Later, she was diagnosed with congenital cataracts and a heart disease (atrial septal defect). The child was held karyotyping, which detected the 18p- syndrome ((46 XX, del (18)(p 11.1; p 11.32)).

At the age of 3 months she was diagnosed with hypopituitarism: secondary hypothyroidism, secondary adrenal insufficiency (Table 2). Brain MRI showed empty sella syndrome. Therapy with levothyroxine (6,25 mkg/kg/day) and hydrocortisone (10mg/m²) has been started, what led to normoglycemia and reduction of cholestasis. At the age of 2 y the girl was diagnosed with growth hormone deficiency.(SDS height=-8,2, bone age-8 months). Growth hormone therapy was started at the age of 3 years, and had a good effect (Pic.1,2).

A girl has a strong psychomotor retardation.

Picture 1. The patient at the age of 1 month and 4,5 years



1 month of life

4,5 year of life

Table 1. Biochemical assessment

Day of life	AST (U/L)	ALT (U/L)	Direct bilirubin (mg/dL)	Total bilirubin (mg/dL)	GGT (U/L)	Cholesterol (mmol/L)	Triglycerides (mmol/L)
26	158 (5-42)	65 (5-40)	41,7 (0-5,1)	222,2 (3,4-17,1)	238 (12-271)	4,1 (1,6-3)	2,57 (0,34-1,6)

Table 2. Hormonal assessment

Day of life	ACTH (pg/ml)	Cortisol (nmol/L)	TSH (mIU/L)	ft4(pmol/L)
62	6,05	0,00	1,6	7,37

CONCLUSIONS

In our case the patient had typical features of the 18p- syndrome with breakpoint in the centromeric region. Further research and monitoring of patients are needed to determine the prognosis of the disease.

References

1. Catherine Turleau Monosomy 18p Review Orphanet Journal of Rare Diseases 2008,3:4
2. Hasi-Zogai M, Sebald C, Heard P, Carter E, Soileau B, Hill A, Rupert D, Perry B. Atkinson S, O'Donnell L, Gelgond J, Lancaster J, Fox PT, Hale DE, Cody JD. A Review of 18p Deletions. American Journal of Medical Genetics Part C (Seminars in Medical Genetics) 169C:251–264 (2015).

Picture 2. Growth data

