Case report: Neonatal McCune-Albright syndrome with juvenile ovarian granulosa cell tumor in a 4 months old girl


(1) Altona Children’s Hospital, Hamburg, Germany
(2) Institute of Pathology, University Medical Center Hamburg-Eppendorf, Hamburg, Germany
(3) Department of Pediatrics, University Medical Center Hamburg-Eppendorf, Hamburg, Germany

Introduction: McCune-Albright syndrome (MAS) is a rare disease resulting from a somatic activating mutation of GNAS1 encoding the Gs-alfa subunit of the G-protein coupled membrane receptor responsible for multiple hormonal signaling cascades leading to the classical trias: polyostotic fibrous dysplasia, café-au-lait hyperpigmentation and GnRh independent precocious puberty. Early manifestation is accompanied by multiple organ involvement and may lead to ACTH-independent hypercortisolism, hyperthyroidism, cardiac alterations, hepatopathy and GH-Excess in addition to the classical trias.

History:
- 2. child, uneventful pregnancy, delivery at 37 weeks of gestational age, birth weight 3505 g, length 52 cm, head circumference 34 cm
- Polydipsia since birth (up to 2 l/day at age of 5 weeks)
- At the age of 5 weeks: weight 4000 g, length 50 cm, h.c. 34 cm
- Further investigation because of heart murmur

Clinical findings and course:
- Hypertrophic obstructive cardiomyopathy (LVEDD 14 mm),
- Glucosuria and hyperglycemia
- Nephrocalcinosis
- Polyostotic fibrous dysplasia
- Autonomous ovarian cysts
- Glucocorticoid excess (free Cortisol 24 h urine: 352 µg/d [n < 70 µg/m² body surface], ACTH <5 µg/l, Cortisol 15:00 338– 370 µg/ml without diurnal rhythm)

Conclusion:
Early manifestation of MAS due to activating GNAS mutation is accompanied by multiple organ involvement including autonomous ovarian cysts.
We present a case with hypercortisolism due to neonatal MAS and effective treatment with metyrapone, who developed juvenile granulosa cell tumor.
It has to be assumed, that the activating GNAS mutation, high estrogen levels and high androgen levels due to side effects of metyrapone caused rapid tumor development.
This has to be taken into account, when making the decision how to treat hypercortisolism in patients with neonatal MAS.