NEONATAL CUSHING SYNDROME ASSOCIATED WITH MCCUNE ALBRIGHT SYNDROME PRESENTING WITH DIABETES MELLITUS

Y UNSAL[1], E N GONC[1], IR USER[2], HIZARCIOGLU H.[3] B GULHAN[4], S EKINCI[2], KARAGOZ T[5] Z A OZON[1]

1. Hacettepe University Faculty of Medicine, Department of Pediatrics, Division of Pediatric Endocrinology, Ankara, Turkey.
2. Hacettepe University Faculty of Medicine, Department of Pediatric Surgery, Ankara, Turkey
3. Hacettepe University Faculty of Medicine, Department of Pediatrics, Division of Pediatric Gastroenterology, Ankara, Turkey
4. Hacettepe University Faculty of Medicine, Department of Pediatrics, Division of Pediatric Nephrology, Ankara, Turkey
5. Hacettepe University Faculty of Medicine, Department of Pediatrics, Division of Pediatric Cardiology, Ankara, Turkey

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INTRODUCTION

- **Neonatal McCune Albright syndrome (MAS) is very rare; presentation is more severe and widespread with multiple organ involvement**
- **Hyperglycemia is very rare at presentation**
- **Outcome of neonatal MAS is usually unfavorable with increased mortality.**

CASE PRESENTATION

- 11 day-old girl was admitted for weight loss and poor well-being.
- History of
  - Respiratory distress
  - Elevated liver enzymes
    - ALT: 2376 U/L; AST: 875 U/L
  - Neonatal sepsis
    - Intravenous antibiotic regimen on first day of life

- Prenatal tests and USG: normal
- Natal:
  - G2P2, 38 weeks of gestation, 2340 g
- Consanguinty and relevant family history were not present.

PHYSICAL EXAMINATION

- **Weight: 2315 g (-2.66 SDS)**
- **Head circumference: 32,6 cm (-1.88 SDS)**
- **BP: 70/30 mmHg**

- **Round facies, elongated philtrum, retromicrognatia**
- **Hyperpigmented macules**
- **Hypertrichosis**

LABORATORY ON ADMISSION

- 11-16th days
  - Breast-fed neonate
  - **Persistent hyperglycemia (210 mg/dl)**
  - Insulin: 18.10 µIU/ml
  - 0.5 units of subcutaneous NPH insulin (3x)

ALT	**539 U/L**
AST	**134 U/L**
Creatinin	0,5 mg/dl
BUN	**53,27 mg/dl**
Na	**126 mEq/L**
K	**3.24 mg/dl**

	Value	Normal range
8 AM ACTH (pg/ml)	5.3	(0-46)
8 AM cortisol (µg/dl)	225.68	(6.7-22.6)
24 hour UFC(µg/day)	**1129**	(4-56)
1 mg DST (cortisol) (µg/dl)	102	(6.7-22.6)

- Hyponatremia and hypokalemia regressed
- **Liver enzymes and renal function tests were elevated.**

20th day

- Hypertension (110/90 mmHg)
- Spironolactone (2 mg/kg/day)
- Nifedipin (0.5 mg/kg/day)

Café au lait macules + **Cushing Syndrome** → **McCune Albright Syndrome**

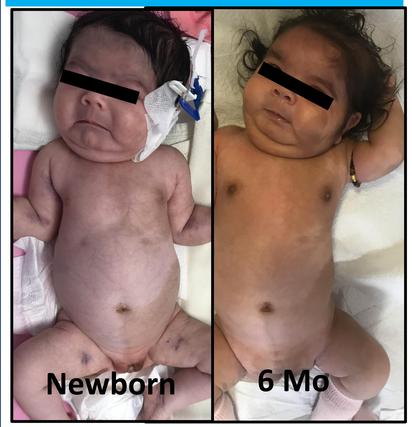
- Euthyroid
- Precoccious puberty at 12 months
- FD at 10 months
- Growth hormone excess ø
- Abdominal USG: Bilateral nodular adrenal hyperplasia

Metyrapone (300 mg/m2/day)

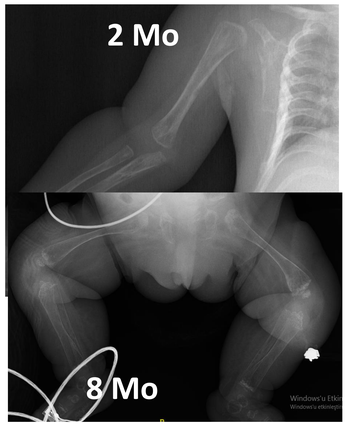
Insulin ceased

Antihypertensives ceased

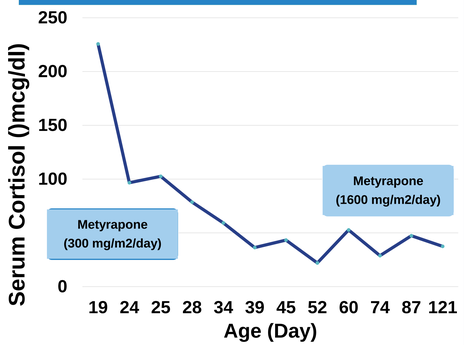
9-MONTH OLD



OSTEOPOROSIS

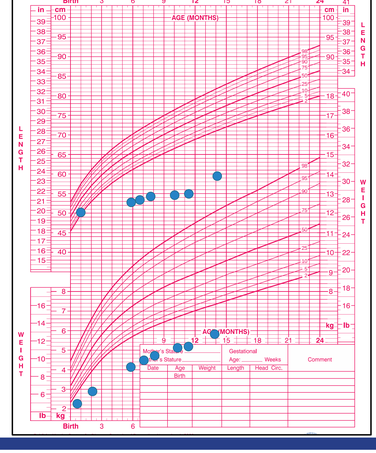


HYPERCORTISOLISM



CARDIAC HYPERTROPHY

GROWTH RETARDATION



10-MONTH OLD

THREE QUARTERS ADRENALECTOMY

Adrenal DNA Sequencing analysis
***GNAS* Exon 8, missense heterozygous pathogenic variant**

CONCLUSION

- **Neonatal MAS has a unique phenotype.**
- **Severity of initial hypercortisolemia is a negative prognostic factor and early adrenalectomy is usually indicated.**
- **Three quarters adrenalectomy is an effective treatment for CS due to MAS.**

CONTACT INFORMATION

Yagmur Unsal, MD
Department of Pediatrics
Division of Pediatric Endocrinology
Hacettepe University Faculty of Medicine,
Ankara, 06230, TURKEY
Phone: +905367950760
Email: yagmurunsal@yahoo.com