Severe Hyponatremia and Repeated Intestinal Resections for Intestinal Dysmotility Mimicking Congenital Aganglionic Megacolon Due to Delay in the Diagnosis of Congenital Hypothyroidism

Gonul Buyukyilmaz[1], Demet Baltu[2], Tutku Soyer[3], Murat Tanyıldız[2], Huseyin Demirbilek[1]

[1] Hacettepe University, Division of Pediatric Endocrinology, Ankara, Turkey
[2] Hacettepe University, Division of Pediatric Intensive Care, Ankara, Turkey
[3] Hacettepe University, Department of Pediatric Surgery, Ankara, Turkey

BACKGRAUND

Congenital hypothyroidism (CH) may presents with non-specific signs and symptoms though majority of infants can be asymptomatic. Therefore, underestimation and delay in diagnosis may results in severe complications. Herein, we report delay in the diagnosis of CH in a female infant, who developed severe neurodevelopmental delay, severe hyponatremia and abdominal distention mimicking congenital aganglionic megacolon which required repeated surgery and releated complications

CASE REPORT

Born after 40 weeks uneventful gestation
- Birth weight was 4000 gr
- Developed prolonged neonatal jaundice
- Treated with phototherapy and phenobarbital

1 month old
- Vomitting, abdominal distention and poor feeding
- Diagnosis of intestinal obstruction due to congenital aganglionic megacolon was considered
- Surgical resection and reanastomosis performed

1-5 months
- Complaints had not been resolved thereby required subsequent six different operations
- 5 months old referred our hospital

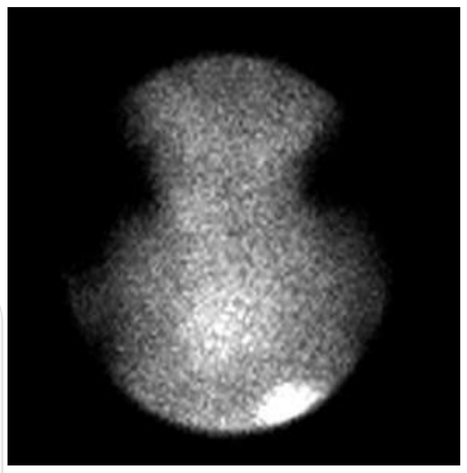
5 months old
- Weight: 3800 gr (<3 pc)
- Anterior fontanelle: 3*3 cm
- Dried and crumped skin
- Abdominal distention
- Neurodevelopmental delay.

•Laboratory investigation at admission:
•**Na :132 mEq/L,**
•**K:3.7 mEq/L,**
•Other tests were normal

2th day of hospitalization
- Developed mild hyponatremia, (**Na :125 mEq/L, K:4.6 mEq/L)**
- No signs of volume expansion or depletion, vomiting, diarrhea or gastrostomy tube loss
- Corrected with saline replacement
- Histopathologic evaluation of previously resected intestinal specimens revealed a normal ganglion cell including colon samples.

3th day of hospitalization
- **Free T4 (FT4): 0.4 pmol/L (7.8-14.4) and TSH: >100 µIU/ml (0.34-5.6)**
- L-thyroxin therapy a daily dose of 50 µg (14.2 µg/kg/day) was started which than tapered to a daily dose of 37.5 µg (10.7 µg/kg/day) on day 4
- Thyroid imaging using ^{99m}Tc-pertechnetate thyroid scan revealed thyroid agenesis (Figure 1)

11th day of hospitalization
- Poor feeding, vomiting, abdominal distention, and respiratory distress
- Laboratory investigations:
  - Severe hyponatremia with normal potassium level (**Na :106 mEq/L, K:4.3 mEq/L**)
  - **FT4: 2.73 pmol/L (7.8-14.4) and TSH >100 µIU/ml (n: 0.34- 5.6)**
- L-thyroxin dose was increased to 50 µg per day
- Infusion of hypertonic saline and subsequent replacement of sodium deficit was commenced



•**Figure 1**. Thyroid agenesis in ^{99m}Tc scan

13th day of hospitalization
- **Plasma sodium level was 120 mEq/L**
- Normal renin and aldosterone levels in course of hyponatremia (Table 1)
- FT4 were stil at hypothyroid level with elevated TSH >100 µIU/ml
- To achieve euthyroid state L-thyroxin dose was increased up to 100 µg/day which was tappered after attaining a normal FT4 level.
- After achievement of euthyroid state plasma sodium level rised to normal ranges and remained stable with no requirement of sodium replacement

Table 1. Presenting and follow up biochemical and hormonal characteristics of patient

	Day 1	Day 2	Day 3	Day 6	Day 11	Day 12	Day 13	Day 16
Plasma Na (mEq/L)	132	125	126	134	106	117	120	136
K (mEq/L)	3.74	4.6	4.4	4,9	4.3	3.9	4.1	5,1
Cl (mEq/L)	103	96	91	108	80	93	95	110
Urine (Na mEq/L)		119					147	
Uric acid (mg/dL)	1.57			2.2	0.43	0.82	0.94	1,55
Osmolality							245	
TSH (µIU/ml)			>100		>100		>100	11
FT4 (pmol/L) (N:7.86-14.5)			0.4		2.73		4.2	33.4
Renin (pg/mL) (N: 2,7-16,5)							5.56	
Aldosterone (pg/mL) (N:10-160)							142	

CONCLUSION

Since presenting symptoms are variable and non-specific, for prompt diagnosis and immediate treatment, congenital hypotyhroidism should be kept in mind in the differential diagnosis of neonates with persistent abdominal distention mimicking aganglionic megacolon and severe hyponatremia of unknown origin.

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