TITLE

Clinical, biochemical and echocardiographic evaluation of patients with congenital rickets due to maternal vitamin D deficiency

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OBJECTIVES

- Vitamin deficient (VDD) rickets can manifest with skeletal (hypocalcemia, hypophosphatemia, elevated serum alkaline phosphatase and defective bone mineralization) and extra-skeletal findings.
- There are certain number of case reports and limited number of small scale studies reporting dilated cardiomyopathy due to VDD rickets.
- There is no large scale studies evaluating the cardiac functions in neonates with VDD rickets.
- The aim of the present study is to evaluate the clinical, biochemical and echocardiographic features of neonates with congenital rickets due to maternal vitamin D deficiency.

METHODS

- Pprospective cross-sectional observational study
- 148 neonates with the diagnosis of VDD rickets were recruited.
- Serum calcium, magnesium, albumin, phosphorus, alkaline phosphatase, parathyroid hormone (PTH), and 25(OH)D3 was measured.
- A low serum calcium elevated PTH and low 25(OH)D3 was defined as VDD rickets.
- Presentation at postnatal first month was considered as congenital rickets.
- Echocardiographic evaluation was performed to all patients.
- Echocardiographic measurement was assessed according to American Echocardiography Association Pediatric Echocardiography guideline.
- Vitamin D3 and oral calcium were administered to all patients,
- A biochemical recovery was achieved in all patients.
- Following remission of clinical and biochemical VDD rickets, a control echocardiography was performed.
- Statistical analysis were perfomed using SPSS for Windows v.22

RESULTS

- The study included 148 cases (95 male).
- 119 out of 148 (80.4%) were presented at the first 72 hours.
- The presenting laboratory features for VDD rickets, echocardiographic parameters for evaluation of dilated cardiomyopathy at presentation and after treatment were shown in Table 1.
- In the echocardiographic evaluation, none of patients had dilated cardiomyopathy.

- There was not a statistically significant difference between the echocardiographic parameters before and after remission of VDD rickets.
- All of the mothers were also vitamin D deficient.

Table 1. Biochemical and echocardiographic findings of 148 neonates with VDD rickets

Biochemical features	Pretreatment Mean±SD (n=148)	Post-treatment Mean±SD (n=148)	P value
Calcium (mg/dl)	7.1±0.5	10.0±0.7	<0,001
Phosphorous (mg/dl)	6.3±1.2	6.1±1.0	0,1
ALP (U/L)	224.0±82.7	314.0±134.5	<0,001
PTH (pg/ml)	133.6.±79.5	44.5±41.9	<0,001
Mg (mg/dl)	1.8±0.2	2.0±0.3	0,07
25(OH)D3 (ng/ml)	5.5±2.4	28.5±14.9	<0,001
Maternal 25(OH)D3 (ng/ml)	7.1±3.5		
Echocardiographic findings			
EF (%)	69.3± 6.1	70.6± 5.9	>0.05
SF (%)	37.8±4.3	38.2±4.0	>0.05
LVEDd (cm)	2.02±0.16	1.94±0.24	>0.05
LVESd (cm)	1.48±0.12	1.49±0.14	>0.05

EF: Ejection fraction, SF: Shortening fraction, LVEDd: Left ventricle end-diastolic diameter, LVESd: Left ventricle end systolic diameter

CONCLUSIONS

To the best of our knowledge, in this largest cohort of patients with congenital rickets due to maternal vitamin D deficiency, echocardiographic evaluation did not show dilated cardiomyopathy in none of 148 cases, despite all presented with hypocalcemia and vitamin D deficiency. In addition, cardiac function did not change with remission of VDD rickets. Absence of dilated cardiomyopathy can be attributed to early diagnosis and lack of prolonged exposure to the hypocalcemia. However, the exact underlying aetiology should be extensively investigated in all cases with dilated cardiomyopathy even in cases with hypocalcemia and VDD rickets.