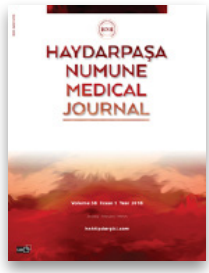
HAYDARPAŞA NUMUNE MEDICAL JOURNAL

DOI: 10.14744/hnhj.2020.79847
Haydarpasa Numune Med J 2020;60(3):284–287



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ORIGINAL ARTICLE

Evaluation of the Patients with Congenital Rickets

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Abstract

Introduction: The present study aims to evaluate the clinical findings and biochemical properties of the patients who were followed up in the neonatal intensive care unit due to congenital rickets.

Methods: The data of patients who were followed up in our neonatal intensive care unit between March 2015 and March 2020 due to congenital rickets were analyzed retrospectively from the patient's files and registration forms. The findings of physical examination and biochemical tests of the patients were recorded at the time of admission.

Results: Six patients were included in this study. Four of the patients were girls. The mean age of application was 19±6 days. All patients were admitted with convulsion. The average calcium at the first application was 5.5±0.7 mg/dL; ionized calcium 0.7±0.02 mmol/L; phosphorus 6.9±1.2 mg/dL; alkaline phosphatase 747±148 U/L; parathyroid hormone 265±40 pg/mL and mean 25-OH vitamin D 3.4±0.4 ng/mL. All cases were hospitalized and treated and diagnosed with congenital rickets.

Discussion and Conclusion: Despite vitamin D prophylaxis, rickets can still be seen in our country. With this study, we aimed to draw attention to the significance of vitamin D prophylaxis.

Keywords: Congenital rickets; hypocalcemia; Vitamin D.

To prevent the development of rickets due to vitamin D deficiency in Turkey, daily doses of 400 UI vitamin D are administered to all newborn babies from the 15th day of birth by the Ministry of Health since 2005[1]. In addition, to prevent maternal/perinatal vitamin D deficiency, vitamin D supplementation (1200 U/day) has been recommended to every pregnant (except in cases where vitamin D will not be applied) since 2011[2]. However, despite all these approaches, severe maternal vitamin D deficiency continues to be a problem frequently seen in Turkey. This is an important risk factor for congenital rickets[3–5]. The present study aims to determine the clinical and biochemical properties of patients who received inpatient treatment in the neonatal intensive care unit with the diagnosis of congenital rickets and to draw attention to the importance of vitamin D prophylaxis once again.

Materials and Methods

In this study, the data of the patients who were followed up in our neonatal intensive care unit between March 2015 and March 2020 due to congenital rickets were analyzed retrospectively from patient files and patient registration forms. Physical examination findings and biochemical tests of the patients were recorded at the time of the admission. Ethics committee approval was received for this study (No: 2019-201).

Physical examinations of the patients were performed on admission. Clinical findings were recorded. Blood calcium (Ca), phosphorus (P), magnesium (Mg), alkaline phosphatase (ALP), 25-OH vitamin D, parathyroid hormone (PTH) levels of the patients and their mothers were mea-

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Submitted Date (Başvuru Tarihi): 21.04.2020 **Accepted Date (Kabul Tarihi):** 09.05.2020

sured. A 25-OH vitamin D level of <12 ng/mL was considered to be a deficiency, 12-20 ng/mL to be insufficiency, and 20-100 ng/mL to be normal.

Statistical Evaluation

The data obtained were analyzed using the statistics program (SPSS) (Version 17, Chicago, IL, USA) prepared for social sciences. Descriptive statistics (mean, standard deviation, median value, minimum, maximum, number and percentile) were given for the variables in this study.

Results

Between March 2015 and March 2020, six patients were admitted to our neonatal intensive care unit with the diagnosis of congenital rickets. Four of these patients were female, and two were male. The mean age of the patients was 19±6 days. All patients applied with the complaints of convulsions. All patients were term except one case. There was a wide fontanelle in two patients and an ankle enlargement in one patient. No other features were detected in the physical examination of the patients. The clinical features of the cases are presented in Table 1.

When the biochemical findings of the patients were examined, mean calcium (5.5±0.7 mg/dL); albumin (4.2±0.3 g/dL), ionized calcium (0.7±0.02 mmol/L); phosphorus (6.9±1.2 mg/dL); alkaline phosphatase (747±148 U/L); parathyroid hormone (265±40 pg/mL) and 25-OH vitamin D (3.4±0.4 ng/mL) values were measured as indicated. No radiological findings other than increased metaphyseal width were detected on the radiographs of the patients. The biochemical findings of the cases are presented in Table 2.

When the biochemical findings of the mothers of the patients were examined, the mean calcium (7.9±1 mg/dL), phosphorus (3.7±0.7 mg/dL); alkaline phosphatase (189±84 U/L), parathyroid hormone (214±103 pg/mL) and 25-OH vitamin D (4.8±2.3 ng/mL) values were determined as indicated. None of the mothers used vitamin D during pregnancy. The biochemical findings of the mothers of the cases are presented in Table 3.

Discussion

Congenital rickets is the term given to the fetus born with the clinical features of rickets, but newborns that do not have prominent clinical features but show the biochemical findings of rickets can also be considered as congenital rickets[6]. Placental calcium transfer is thought to protect the fetus from rickets[6]. Therefore, clinical and radiological findings may be uncertain in cases of rickets seen in the neonatal period, and the disease may occur only with hypocalcemia or hypocalcemic seizure as in our cohort[6, 7].

Table 1. Clinical findings of the patients

No	Gender	Gestational age	Birth weight (g)	Age at admission (days)	Admission complaint	Nutrition	Vitamin D prophylaxis
1	Female	Term	3410	12	Convulsion	BM	No
2	Male	Term	3240	25	Convulsion	AS	Irregular
3	Male	Preterm	2200	28	Convulsion	AS+ formula	Irregular
4	Female	Term	3010	17	Convulsion	AS+formula	No
5	Female	Term	2950	21	Convulsion	BM	Irregular
6	Female	Term	2870	14	Convulsion	AS	No

Table 2. Laboratory findings of the patients

No	Calcium (mg/dL)	Phosphorus (mg/dL)	Magnesium (mg/dL)	Albumin (g/dL)	Ionized calcium (mmol/L)	Alkaline phosphatase (U/L)	PTH (pg/mL)	25-OH vitamin D (mg/dL)
1	5.8	5.6	1.85	4.7	0.75	640	318	<3
2	6.1	9.1	2.07	4.1	0.77	605	281	3.91
3	5.5	6.8	1.93	4.0	0.71	853	239	<3
4	4.8	6.1	1.55	4.5	0.73	678	224	3.0
5	4.5	7.2	1.56	3.8	0.72	995	302	<3
6	6.3	6.6	2.16	4.6	0.78	711	229	3.53

Normal values: Calcium; 8.0-10.8 mg/dL phosphorus 4.8-8.2 mg/dL, magnesium;1.7-2.7 mg/dL, Alkaline phosphatase; 150-420 U/L, PTH;15-87 pg/mL, 25-OH vitamin D; 20-100ng/mL: BM, breast milk.

Table 3. Laboratory findings of the mothers of the patients

No	Calcium (mg/dL)	Phosphorus (mg/dL)	Alkaline Phosphatase (U/L)	PTH (pg/mL)	25-OH vitamin D (mg/dL)	Use of vitamin D during pregnancy
1	6.7	5	344	397	<3	No
2	8.4	3.9	130	190	3.3	No
3	8.5	3.1	208	212	9.1	No
4	8.7	3.3	103	106	4.0	No
5	8.9	3.0	166	133	3.9	No
6	6.8	4.1	184	248	3.9	No

Normal values: Calcium; 8.0-10.8 mg/dL phosphorus 4.8-8.2 mg/dL, magnesium;1.7-2.7 mg/dL, Alkaline phosphatase; 150-420 U/L, PTH;15-87 pg/mL, 25-OH vitamin D; 20-100ng/mL.

In congenital rickets, hypotonia, severe tremor, serum alkaline phosphatase elevation, 25-OH vitamin D deficiency, secondary hyperparathyroidism, hypocalcemia and resistant convulsions due to hypocalcemia may be seen[8]. In addition, unlike classical rickets, laboratory findings may accompany hypophosphatemia, as well as hypophosphatemia in cases of congenital rickets[7]. All these conditions may cause difficulty in the differential diagnosis.

In this single-center retrospective study, the clinical and laboratory findings of the six patients who were treated in the neonatal intensive care unit for congenital rickets were examined. It is noteworthy that the mothers of all patients had low 25-OH vitamin D levels and did not receive the appropriate doses of vitamin D prophylaxis during pregnancy. Laboratory and radiological findings of all our patients were compatible with congenital rickets. In addition to hypocalcemia, hyperphosphatemia, all patients had increased ALP, and PATHOLOGY, but decreased 25 (OH) vitamin D levels. In addition, mothers of the patients had lower 25 (OH) vitamin D levels.

The only source of vitamin D for the fetus is vitamin D, which passes through the placenta from the mother. A low 25-OH vitamin D level was found in the cord blood of infants of mothers with maternal vitamin D deficiency[9, 10]. In addition, it has been seen in many studies conducted to date that maternal vitamin D deficiency is the most important risk factor for vitamin D deficiency in the neonatal period and early infancy[11]. This increases the risk of congenital rickets in babies born to mothers with low 25-OH vitamin D levels.

As in our study, congenital rickets may present with life-threatening hypocalcemic seizures. Congenital rickets are a very important preventable public health problem concerning causing permanent complications. The requirement and dosage of vitamin D supplementation for pregnant women may vary according to countries and regions. Despite all the developments in the field of public health in our country, it is seen that clinical problems related to vitamin D deficiency remain an important issue today.

Given that clinical and radiological findings of congenital rickets may not appear over time, it is also important to evaluate children who are admitted to the hospital concerning rickets findings, especially in regions with low socioeconomic status[12].

In conclusion, our experience with these six patients has shown that maternal vitamin D deficiency is still seen in our country and remains important as a severe health problem. Thus, to prevent maternal/perinatal vitamin D deficiency, appropriate doses of vitamin D supplementation are very important for every pregnant woman.

Ethics Committee Approval: Istanbul Medipol University Ethics Committee, No: 2019-201.

Peer-review: Externally peer-reviewed.

Conflict of Interest: None declared.

Financial Disclosure: The authors declared that this study received no financial support.

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