

## Congenital Rickets: Report of Four Cases

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### Abstract

#### Introduction:

Vitamin D deficiency and rickets continue to be health problems in developing countries and most of the infants with congenital rickets may present with hypocalcemic seizure.

#### Case Report:

In this article, the report on four infants who presented with hypocalcemic seizures but subsequently were found to have congenital rickets is presented. All of them had hypocalcaemia and low level of serum 25-hydroxy vitamin D. Their mothers had not received vitamin D supplementation during pregnancy and so evidence of vitamin D deficiency was presented.

#### Conclusion:

Although current vitamin D supplementation guidelines for infants was effective in prevention of rickets in Iranian children, it is necessary to evaluate women before pregnancy to prevent this entity. Also infants without vitamin D supplementation therapy who present with seizures during the first 6 months of age should undergo biochemical and other investigations for rickets.

#### Keywords:

Congenital rickets, Vitamin D deficiency, Hypocalcemia, Seizure.

### Introduction

Nutritional rickets is present more commonly in Africa, the Indian subcontinent, Asia, Latin America and the Middle East (1). In developing countries most women at childbearing age have vitamin D deficiency which is an important risk factor for congenital rickets (2-7). Severe vitamin D deficiency (<20 mol/L) has been detected in

46 to 80% of pregnant women and nursing mothers in different regions in Turkey, and their dietary Vitamin D intake was below the recommended dose (185±117/day) (3,8).

Also vitamin D deficiency is reported in Tehran, capital of Iran as a frequent problem. According to this study the prevalence of severe and moderate Vitamin D deficiency in women below 50 years old were 0.3-10.6% and 32.8-49.8% respectively (9).

Rickets in infants and children caused by an inadequate vitamin D diet was common in the industrialized world until about the middle of the last century. Improved public health measures and the fortification of cow's milk and infant formulas with

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vitamin D resulted in the virtual elimination of the disease in such countries (12). The current report is an experience in last year (2011) with cases of congenital rickets who presented with hypocalcemic seizures.

If rickets presented in newborn period or in early infancy (<6months of age) it is called as congenital rickets (10). Congenital rickets is characterized by low 25OH vitamin D levels in both infant and mother and it differs

from vitamin D deficient rickets in older children because of phosphate level and probably radiologic findings (11).

**Case Report**

The chemical and biochemical findings of the patients who were all seen by the authors are summarized in tables 1 and 2.

**Table1:** Summary of main clinical and radiological findings in four infants with congenital rickets.

| Patient          | Infant's data |         |            |           |               | Mother's data |         |            |               |
|------------------|---------------|---------|------------|-----------|---------------|---------------|---------|------------|---------------|
|                  | Ca mg/dl      | P mg/dl | ALP Unit/L | PTH Pg/mL | 25(OH)D ng/ml | Ca mg/dl      | P mg/dl | ALP Unit/L | 25(OH)D ng/ml |
| 1                | 5.7           | 6       | 1270       | 172       | 4             | 8.9           | 4.1     | 225        | 7.5           |
| 2                | 4.5           | 3.8     | 1108       | 85        | 7             | 10.2          | 4       | 280        | 4.5           |
| 3                | 6             | 6.8     | 1530       | 43        | 10            | 9.3           | 3.5     | 345        | 6             |
| 4                | 5.5           | 3.2     | 1729       | 369       | 3             | 9.3           | 4.9     | 204        | 3.7           |
| Reference values | 8.5-10.5      | 4.5-6.5 | 180-1200   | 10-65     | 20-100        | 8.5-10.5      | 3-5     | 30-240     | 20-100        |

All patients were treated with calcium and 1000-10000 IU vitamin D per oral daily and all of them were cured clinically and biochemically in several weeks.

**Case 1**

A two – month – old infant was admitted in emergency room due to recurrent seizures from 3 days ago. In the last day convulsions repeated every one hour. Initial Laboratory study revealed a serum total calcium 5.7 mg/dl (8.5-10.5), albumin 3.5 gram/dl, and ALP 1270 IU (180-1200).

After calcium administration, serum calcium changed to 8.3 mg/dl after 48 hours, and then 10000 IU vitamin D daily for 6 weeks continued. In outpatients follow up he was found to be a well developed and well nourished normal baby.

**Case 2**

A 3-month- old male that referred to the center because of frequent seizures and detection of serum calcium 4.5 mg/dL (8.5 - 10.5). His 20- year- old healthy mother had not taken vitamin D supplements during pregnancy.

He was admitted because of frequent seizures and fever during the past 48 hours. Treatment with intravenous calcium gluconate started and serum calcium raised to 7.3 mg/dL after 48 hours. Treatment with oral calcium and vitamin D3(10000 iu/day) continued for 6 weeks.

**Case 3**

28-day-old neonate who was admitted in hospital because of frequent seizures but his parents refused more evaluation and left the hospital was admitted again 18 days later due to recurrent convulsion in neurology ward and when hypocalcemia was detected, he was referred to endocrine ward. His parents were relatives and his mother had not gotten vitamin D in pregnancy.

**Case 4**

She was a 5.5- month- old infant who referred to the hospital because of frequent seizures and after initial evaluation, hypocalcemia was observed .she was the only infant whose signs of rickets were typically present in radiographs so diagnosis of rickets was established and treatment began.

**Table2:** Summary of biochemical findings in four infants with congenital rickets and their mothers.

| Patient | Sex | Age (m0) | Gestational age | Delivery | Birth weight | Now weight | Clinical manifestation                      | Feeding                  | Radiographic findings                 |
|---------|-----|----------|-----------------|----------|--------------|------------|---|--------------------------|---------------------------------------|
| 1       | M   | 2        | term            | NVD      | 2500         | 4600       | Recurrent generalized tonic colonic seizure | Exclusive breast feeding | osteopenia                            |
| 2       | M   | 3        | term            | NVD      | 2500         | 6000       | Recurrent generalized tonic colonic seizure | Exclusive breast feeding | osteopenia                            |
| 3       | M   | 1.5      | term            | NVD      | 3200         | 4500       | Recurrent generalized tonic colonic seizure | Exclusive breast feeding | osteopenia                            |
| 4       | F   | 5.5      | term            | NVD      | 3500         | 6000       | Recurrent generalized tonic colonic seizure | Exclusive breast feeding | Flaring, cupping, fraying, osteopenia |
| patient | Sex | Age (m0) | Gestational age | delivery | Birth weight | Now weight | Clinical manifestation                      | feeding                  | Radiographic findings                 |
| 1       | M   | 2        | term            | NVD      | 2500         | 4600       | Recurrent generalized tonic colonic seizure | Exclusive breast feeding | osteopenia                            |
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| 3       | M   | 1.5      | term            | NVD      | 3200         | 4500       | Recurrent generalized tonic colonic seizure | Exclusive breast feeding | osteopenia                            |
| 4       | F   | 5.5      | term            | NVD      | 3500         | 6000       | Recurrent generalized tonic colonic seizure | Exclusive breast feeding | Flaring, cupping, fraying, osteopenia |

Hypocalcemic seizure at presentation, mothers who did not receive vitamin D in pregnancy and also did not give vitamin D to their kids, age of the patients, who were all below 6 months, at the time of admission were common in all of them.

**Discussion**

The only source of vitamin D available in fetus is that which is derived from the mother. Infants of mothers with vitamin D deficiency have low level of 25-hydroxy vitamin D, and characteristically presented with hypocalcaemia seizures (13).

Newborn vitamin D concentrations have been reported to be in the range of 50-70% of the mothers concentrations (14,15). Infants born to mothers with vitamin D deficiency may have retarded growth and large fontanells, craniotabes and wrist ossification centers.

Although vitamin D deficiency is a completely preventable disease Heshmat et

al (2008), in a multi – center study showed high prevalence of moderate to severe vitamin D deficiency in Iran(9).

Also Clinical and biochemical symptoms of vitamin D deficiency in early infancy may be different from those of the classic rickets. Hatun et al (2005), showed that the most common presenting symptom in this age group was seizure and skeletal deformities were minimal (10). Also in the current report patients except number 4 who had symptoms of rickets on wrist x-ray, the others had no specific symptoms of rickets on their radiographs and all of the cases had referred to the hospital because of seizures.

Biochemically serum calcium is low, phosphorus may be low, high or normal, serum ALP is usually high, serum PTH may be high or normal and serum 25(OH) vitamin D is low in infants and mothers in the cases (10). Innes et al (1999), reported high ALP in 3 out of 4 cases, and high level of PTH in all cases (12). Gul (2011) yesiltepe mutlu et al, in Turkey reported that infants with serum 25-OH-D levels lower than 20 ng/ml had significantly higher PTH levels compared with those with vitamin D sufficiency (25-OH-D>20 ng/ml).there was no significant differences in ALP levels between those two groups (16).

In the current study cases serum phosphorus was in normal range in two case and low in the others, ALP was high in three cases and high normal in one case, and serum PTH was high in three cases, and near normal in the other one. Vitamin D status in two cases was severely deficient (<5ng/ml) and in the rest of cases was deficient (<20ng/ml) (17).

According to the current study data, level of vitamin D was appropriately correlated to the level of ALP and PTH. Hatun et al (2005), concluded that maternal vitamin D deficiency is an important risk factor for vitamin D deficiency in early infancy (10).

Cases in the current study presented in February when there was limited sunlight exposure during autumn and winter, in addition to cultural and traditional clothing.

In primary health care in Iran supplementation with 400IU Vitamin D from the 10th day of life were recommended, but it may be ignored by parents most likely because of poor access to health care state.

### Conclusion

The four babies were treated with 1000 – 10000 IU/day vitamin D and they all recovered rapidly from rickets, thus excluding vitamin D-dependent rickets (18,19).

Congenital rickets, which is quite rare in industrialized countries, occurs when there is

severe maternal vitamin D deficiency during pregnancy. Maternal risk factors include poor dietary intake of vitamin D, lack of adequate sun exposure, and closely spaced pregnancies. So Biochemical screening of pregnant Asian women for vitamin D deficiency or osteopenia should be a routine part of antenatal care, and vitamin D supplementation must be made. Also, this article suggests that hypovitaminosis D is a public health problem in IRAN.

Because of high the rate of vitamin D deficiency in Iranian adult population vitamin D supplementation during pregnancy and lactation to avoid congenital and nutritional rickets on early infancy should be emphasized.

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