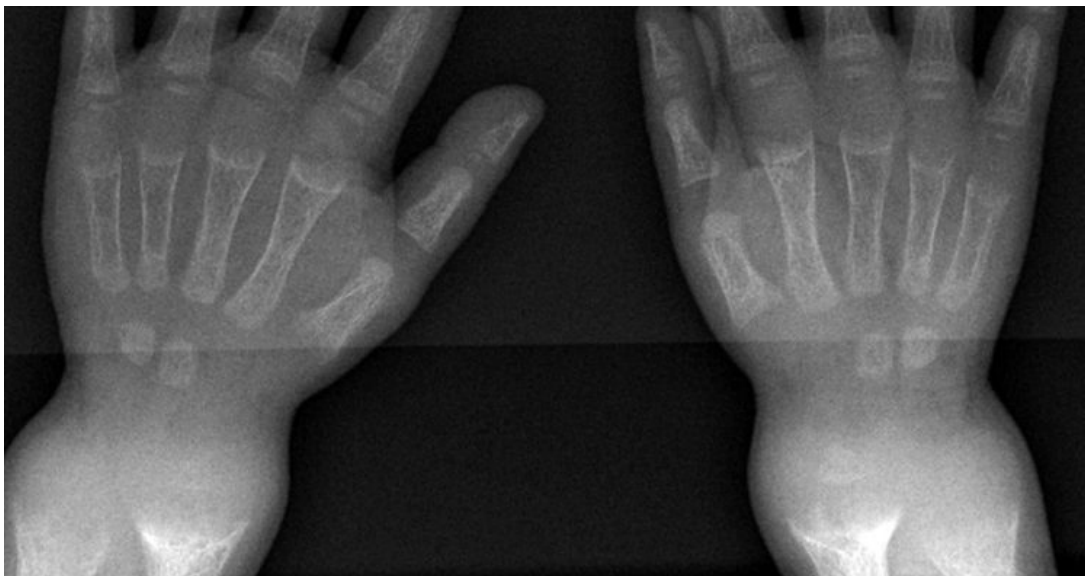


## Rickets (Osteomalacia) — Symptoms and Treatment

[See online here](#)

**Rickets, though not a common disease in the affiliated population, is one of the diseases which affect the growth of children in the developing countries (where the nutritional deficiency is common). Rickets can be characteristically identified using the clinical features and they can be confirmed using biochemical investigation. In this article, we will discuss the various features of rickets in detail.**



### Definition and Epidemiology of Rickets

According to standard textbooks, rickets is defined as the imperfection, distortion and softening of the bones due to **deficiency of vitamin D**. Vitamin D is a fat-soluble vitamin and one of the essential vitamins for the body.

As rickets is mainly caused by the deficiency of vitamin D, and Vitamin D is a hormone which is synthesized on **exposure to sunlight**, the epidemiology of rickets mainly consists of those regions where exposure to sunlight is reduced. The **increase in the growth spurt** especially in puberty is one of the reasons for the manifestation of rickets.

The regions where the diet consists of **inadequate vitamin D and calcium** intake also are linked to the development of rickets.

Though rickets is rare in developed countries, it is frequent in children in **developing countries**. **Poverty** and **inadequate availability of food** are also reasons for the development of rickets in children.

# Etiology and Classification of Rickets

Vitamin D is a fat-soluble hormone responsible for maintaining **serum phosphate and calcium levels**, which in turn are of foremost importance for **bone mineralization**. Exposure to sunlight causes the conversion of vitamin D from its inactive to the active form in the cells of the skin.

The primary reason for **congenital rickets** is the deficiency of vitamin D in the mother. Vitamin D is transported in the form of **25 hydroxyvitamin D** from the mother to the fetus as this form crosses the placenta.

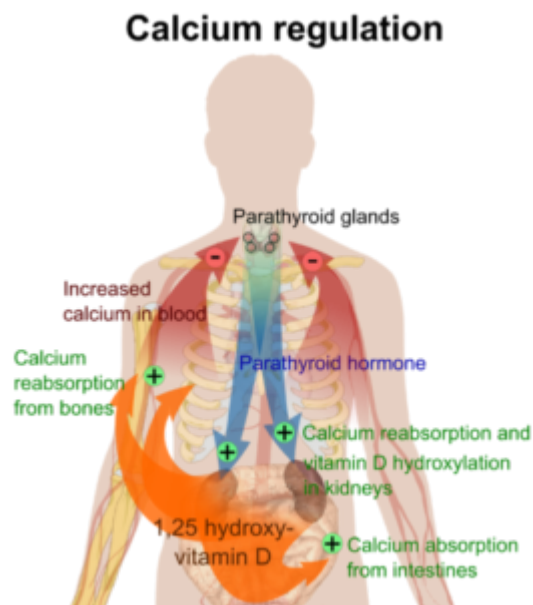


Image: "Overview of calcium regulation." by Mikael Häggström. License: Public Domain

Some of the **risk factors** for the development of vitamin D deficiency in the newborn infant are:

- Deficiency of vitamin D in the mother during pregnancy
- Prolonged breastfeeding without vitamin D supplementation
- Dark skin complexion. It is postulated that northern latitudes have selection for lighter skin that allows UV rays to produce Vitamin D from 7-dehydrocholesterol. Conversely, latitudes near the equator have selection for darker skin that can block the majority of UV radiation to protect from toxic levels of Vitamin D, as well as skin cancer.
- Very low sun exposure

In addition to this, other **gastroenterological interventions** that remove the lower part of the small bowel where Vit D is absorbed and **diseases of the intestine** that impair the normal absorption both decrease the uptake of vitamin D even in the presence of adequate nutritional supplementation.

The primary reason for rickets in children is a deficiency of vitamin D. Vitamin D is synthesized by exposure to sunlight. Insufficient exposure to sunlight and **certain dietary habits** like a diet exclusively based on cereal are biological reasons for the development of rickets in children.

In some cases, there may be a normal level of vitamin D but decrease in activity due to lack of conversion to the active metabolite or resistance of the receptor to the

metabolite. This results in what is known as **calciopenic rickets**.

There is also a **genetic cause for rickets** and it is known as **vitamin D Resistant Rickets (X-linked hypophosphatemia)**. It is also known that it can also occur because of **phosphate deficiency**, which has been reported as the cause for rickets in children.

The broad categories of rickets consist of:

#### **Vitamin D-related rickets**

- Vitamin D deficiency leading to rickets
- Vitamin D dependent rickets
  - Type 1: deficiency of the 1-alpha-hydroxylase enzyme
  - Type 2: a mutation in the calcitriol receptor

#### **Hypocalcemia-related rickets**

- Due to direct hypocalcemia
- Due to **hypoparathyroidism, chronic renal failure** (as the kidneys are responsible for the activation of vitamin D and help in the reabsorption of calcium), or malabsorption

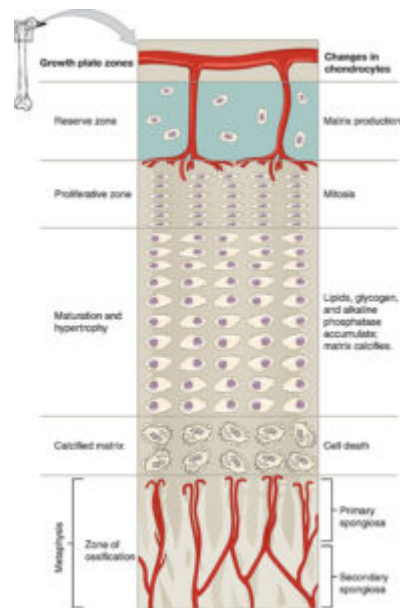
#### **Hypophosphatemia-related rickets**

- Congenital hyperphosphatemia
  - Autosomal dominant hypophosphatemic rickets
  - Autosomal recessive hypophosphatemic rickets
  - Vitamin D-resistant rickets
- Secondary to malabsorption
- Fanconi syndrome, a syndrome of inadequate reabsorption in the proximal renal tubules of the kidney causing loss of phosphate

#### **Rickets secondary to other diseases**

- In addition to this, rickets might arise secondary to other diseases such as **epidermal nevus syndrome** and **McCune Albright syndrome**.

## Pathophysiology and Symptoms of Rickets



**Image:** "Diagram of longitudinal bone growth." by OpenStax College - Anatomy & Physiology, Connexions Web site. <http://cnx.org/content/col11496/1.6/>, Jun 19, 2013. License: [CC BY 3.0](https://creativecommons.org/licenses/by/3.0/)

**Calcium** is absolutely essential for the **mineralization of the chondrocytes**, which are formed in the growth plate. In calcium deficiency, though there occur **proliferation and hypertrophy of chondrocytes**, the concurrent invasion by the vascular tissue and the conversion of the formed chondrocytes into a mineralized structure is deficient.

This results in a **disorganized growth plate** and **expansion of the proliferative zone**. Ultimately, the stability of the bone is severely compromised leading to **bowing** and other characteristic features of rickets.

The symptoms are very characteristic in children.

1. The fontanelles are present at the junction of the skull bones and in the case of rickets, there occurs **delay in the time of closure of fontanelles**. As mineralization of the skull bones is deficient, there is bossing due to the **pressure by the underlying brain** and this is especially prominent in the parietal and frontal regions.
2. Both **Genu varum, as well as Genu valgum**, can occur based on the age at which the rickets manifest. In case of rickets in toddlers, Genu Varum occurs, whereas in older children, Genu valgum occurs.
3. Deformity in the spine (**kyphoscoliosis**) and an increase in the risk of **tetany**
4. The skull bones are soft and this is known as **craniotabes**.
5. The ribs are not strong enough to support the pull which occurs during respiration. This results in the formation of a characteristic sulcus which is present at the lower margin of the chest known as **Harrison sulcus**.
6. The distal **bones broaden** especially the upper arm bones (radius and ulna) and the lower arm bones (tibia).
7. Proliferation of chondrocytes in rickets manifest as the enlargement of the costochondral junction which forms characteristic beading along the chest known as **rachitic rosary** and there also is **widening of the wrists**.
8. The bones are **tender**, there are **problems with the dental architecture**, **greenstick fractures** occur and there is **increased weakness of the muscle**. The growth of the child is affected and disturbed.

# Progression and Complications of Rickets

The progression of rickets leads to the bowing of both the legs with the development of Genu valgum. Growth is also severely stunted due to the occurrence of rickets in the younger age group. Genu valgum is characteristic known as the **knock knees** in which the straightening of the leg leads to the touching of both the legs to one another and the knees are basically angled in.

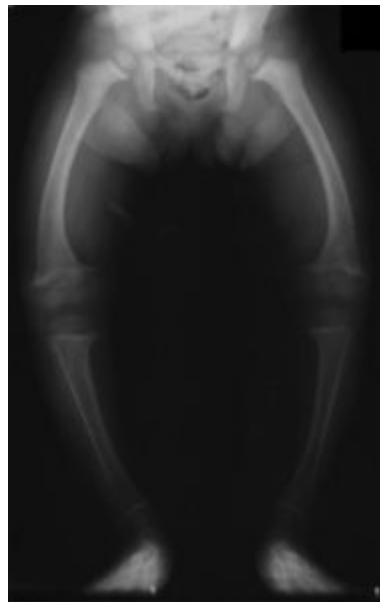
The greatest interference with growth of the child happens when rickets is present in the younger group. This results in the development of **short stature**. Chronic rickets also leads to the development of **osteoarthritis** and other degenerative disorders of the bone. Ultimately, this results in **osteopenia** and a **decrease in the bone mineral density**.

## Diagnosis and Differential Diagnosis of Rickets

A **decrease in the level of calcium and phosphate** is seen in the blood of the patient. **Alkaline phosphatase**, which represents the activity in the bone, is generally **high** in patients with rickets.

The persons suffering from **calciopenic rickets** usually have **elevated parathyroid hormone** and this increase might initially compensate for the decrease of the calcium level. But ultimately, a decrease in the serum calcium levels occur. In addition, **urine calcium/urine creatinine ratio** should be tested.

## Differential diagnosis of rickets



*Image:* "Anteroposterior view of the legs in a 2 year old child with rickets." by The original uploader was Mrich at English Wikipedia - Image created by Michael L. Richardson, M.D. Sept 28th, 2004.  
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- **Achondroplasia** and **metaphyseal chondrodysplasia** characteristically presents similarly with **bilateral bowed legs** but it should be noted that

**inorganic phosphorus** and **PTH hormone levels** would be normal and these would help in the exclusion of these diseases.

- **Renal osteodystrophy:** It is well known that the **kidney** is an important organ where the formation of **1,25 dihydroxy vitamin D** occurs. In addition to this, the kidneys also play a central role in maintaining calcium homeostasis. So in case of **renal insufficiency**, there are concomitant bone diseases characteristically known as renal osteodystrophy. This mimics rickets but can be excluded by means of taking into consideration the **serum creatinine** of the patients (would be elevated).
- **Blount disease:** Disruption of the **proximal tibial physis** (growth plate) on the medial aspect. The radiological features form the key in differentiating.
- Other differential diagnosis include **hypophosphatasia** (the difference is that there would be a low level of alkaline phosphatase), **transient hyperphosphatemia**.

## Treatment of Rickets

As the disease occurs due to the deficiency of vitamin D, calcium and phosphate, the **supplementation of all the three in the diet** form the main part of the treatment. In addition to this, the **exposure to sunlight (ultraviolet B)** is a treatment for this patient (there is another school of thought of recommendation of supplementation instead of the exposure, as increased exposure increases the **risk of skin cancer**). The diet rich sources of vitamin D are **cod liver oil** and **halibut oil**.

In the case of vitamin D deficiency rickets (nutritional rickets), the treatment consists of **ergocalciferol (vitamin D2)** and **cholecalciferol (vitamin D3)**. There are two schools of thoughts on starting the high dose vitamin D or the low dose vitamin D. Vitamin D is generally given 5,000 to 10,000 International Units (IU) and is to be given until the biochemical values are normal after radiological cure occurs for the patient.

If the calcium supplements are not present in the treatment, then there is an **elevation of parathyroid hormone** and it might result in a syndrome known as **hungry bone syndrome**, a rapid 'rebound' recalcification of bones after prolonged hypocalcemia; in order to avoid this, vitamin D supplements need to be given along with calcium.

The recommended dose for infants is 400 IU a day. In Europe, stoss therapy (Single high-dose oral vitamin D3 (stoss) consists of giving 6 lakh international units in a single day for the treatment. The single-day stoss dose should be divided into 4 to 6 doses in the day. The advantages are that there is no issue about compliance (which might be affected in the gradual method). The disadvantage of this therapy is the increased **risk of hypercalcemia**.

In the case of severe deformity, the **orthopedic correction** is required for the deformities. This is especially required for the Genu valgum. Reconstruction surgery is done to straighten the patient's leg.

## Prevention of Rickets

Adequate **exposure to sunlight** holds the key to the prevention of rickets. In order to avoid congenital defects, the mother needs to have sufficient sources of vitamin D from the diet as well as on exposure to the sunlight. A **diet rich in vitamin D** also needs to be taken regularly for prevention.

The **supplementation of vitamin D** and the intake of a diet rich in vitamin D is

especially required in the growing age group of puberty. In the case of children who are exclusively breastfed, the amount of vitamin D present in human milk is very low, especially when babies weight less than 1.5 kg. These are the infants in whom the vitamin D supplements are recommended and the recommendations are given by the US Endocrine Society Clinical Practice and the European Society for Pediatric Gastroenterology.

One of the other innovative steps in preventing the occurrence of rickets is the fortification of milk and cereals with vitamin D.

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