



Australian Paediatric Surveillance Unit  
STUDY PROTOCOL  
Simple Vitamin D Deficiency Rickets

COMMENCING  
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## BACKGROUND

Vitamin D is critical for calcium homeostasis and for mineralization of the skeleton, especially during the growing years. A deficiency in vitamin D leads to rickets and osteomalacia. These effects are associated with pain, fractures, skeletal deformity, growth retardation, dental enamel defects, delayed developmental milestones and, in severe cases, hypocalcaemic tetany and seizures. If not recognized and properly treated, simple vitamin D deficiency may have long-term sequelae. On the other hand, the disease is entirely preventable with simple dietary measures or vitamin D supplementation.

The main site of vitamin D synthesis in the body is in skin exposed to sunshine. In Australia, the majority of infants and children have adequate skin exposure to sunlight for vitamin D synthesis. Despite this, evidence suggests that simple vitamin D deficiency is increasing in Australia (1-6) and other developed countries worldwide (7-11). The literature, and our own experience, suggests that the aetiology of simple vitamin D deficiency rickets over the past decade has been multi-factorial. Children at risk of simple vitamin D deficiency include: a) children born to mothers who are vitamin D deficient (seen most commonly in mothers with dark skin colour who are veiled) (2,5), b) children with dark skin colour, especially if recently immigrated to Australia (5), c) children with a chronic disability/illness that limits mobility and exposure to sunlight (5), d) children who avoid sun exposure and/or use excessive sun screen or veiling (1), e) children with severe allergies and eczema who receive food alternatives with inadequate calcium and vitamin D (12) and f) children with iron deficiency as there is evidence of a physiological interaction between the iron and vitamin D (5).

The aim of this study is to provide unique and current national data on the incidence and aetiology of simple vitamin D deficiency rickets in Australia. By determining the geo-ethnic populations in Australia at most risk of Vitamin D deficiency and documenting the role of recognised risk factors, we will provide data to inform specific health policies for prevention and early identification of simple vitamin D deficiency rickets.

## STUDY OBJECTIVES

1. To estimate the incidence of simple vitamin D deficiency (also known as nutritional rickets) among children living in Australia by identifying all cases newly diagnosed by paediatricians and child health specialists over a two-year period.
2. To obtain demographic and medical information which will assist in: documenting known risk factors for development of disease (poor dietary intake of vitamin D, lack of vitamin D supplementation when indicated, inadequate sun exposure, ethnicity); and evaluating current preventive strategies.
3. To supply data that will inform public health policies for prevention of vitamin D deficiency rickets in children living in Australia.

## CASE DEFINITION AND REPORTING INSTRUCTIONS

*Please report all children aged  $\leq 15$  years of age with rickets secondary to simple Vitamin D deficiency (also known as nutritional rickets) confirmed biochemically and/or radiographically.*

### Biochemical criteria for inclusion as a case of Vitamin D deficiency (nutritional) rickets

1. Low serum 25-hydroxy vitamin D (25OHD)
2. Elevated serum alkaline phosphatase

### Exclusion criteria

1. Vitamin D deficiency rickets associated with underlying disease e.g. fat malabsorption, liver disease and renal insufficiency.
2. Vitamin D deficiency rickets in patients receiving total parenteral nutrition.
3. Vitamin D deficiency rickets secondary to heritable disorders of vitamin D metabolism, including:
  - $1\alpha$ -hydroxylase deficiency (pseudo-vitamin D deficiency rickets)
  - Vitamin D receptor defects (hypocalcaemic vitamin D resistant rickets)
3. Phosphopaenic rickets of any aetiology (where hypophosphatemia is the primary cause of the rickets and not due to calcipenic rickets with secondary hyperparathyroidism).

**When a diagnosis of vitamin D deficient rickets is made the following supplemental tests will ideally be performed prior to treatment (the expected results are indicated in brackets).**

*Note: These results are not essential for reporting a case to the APSU.*

1. Serum calcium and albumin (normal or low)<sup>†</sup>
2. Serum phosphate (normal or low)
3. Serum parathyroid hormone (elevated)

4. X-ray confirmation of rickets at the distal ulna or femoral epiphysis<sup>†</sup>
5. Haemoglobin, mean corpuscular volume and serum Ferritin

† Ionized calcium is also acceptable.

‡ In rare instances, x-ray features of rickets may not be present at diagnosis e.g. if linear growth is arrested (and growth plate activity is blunted) or in the very early phase of the disease when x-ray changes at the growth plate are not yet visible. Thus, although x-ray confirmation of rickets is not a strict diagnostic criterion, it should be obtained during the initial patient evaluation.

#### **Common clinical presentations:**

Children with simple vitamin D deficiency rickets may present with limb deformity, fracture, bone pain, seizures, motor delay, hypotonia, poor growth or respiratory illness. Alternatively, vitamin D deficiency may be found on testing of siblings of an affected child or on routine screening of at risk children. Infants and children at the highest risk of developing simple vitamin D deficiency rickets include a) children born to mothers who are vitamin D deficient (seen most commonly in mothers with dark skin colour who are veiled), b) children with dark skin colour, especially if recently immigrated to Australia, c) children with a chronic disability/illness that limits mobility and exposure to sunlight and d) children who avoid sun exposure and/or excessive use of sun screen or veiling.

#### **FOLLOW-UP OF REPORTED CASES**

A questionnaire requesting further details will be forwarded to clinicians that report a case of Simple Vitamin D Deficiency Rickets to APSU.

#### ***If you have any comments or questions please contact:***

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**This study has been approved by a Human Research Ethics Committee properly constituted under NHMRC guidelines.**