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Implementing Knowledge Based System for Diagnosing and Treating Rickets

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Abstract: Background: The epidemic scourge of rickets in the 19th century was caused by vitamin D deficiency due to inadequate sun exposure and resulted in growth retardation, muscle weakness, skeletal deformities, hypocalcemia, tetany, and seizures. The encouragement of sensible sun exposure and the fortification of milk with vitamin D resulted in almost complete eradication of the disease. Objectives: this paper is going to resolve the exiting problems of rickets by correctly diagnosing and offering the proper treatment. Methods: In this paper, we present an expert system for rickets diagnosis which will helps doctors to explore everything related to the problems of rickets. We look forward to offer simplified answers to most of the rickets disease.

Keywords: Artificial Intelligence, Expert Systems, SL5 Object, Rickets.

1. INTRODUCTION

Rickets is a skeletal disorder that results from a lack of vitamin D, calcium, or phosphate. These nutrients are important for the development of strong, healthy bones. People with rickets can have weak and soft bones, stunted growth, and, in severe cases, skeletal deformities [7, 8] see Figure 1 for details.

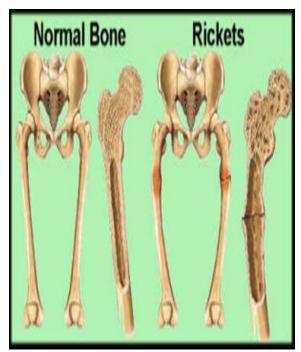


Figure 1: The figure presents Normal Bone and Rickets.

Vitamin D helps your body absorb calcium and phosphate from your intestines. You can get vitamin D from various food products, including milk, eggs, and fish. Your body also produces the vitamin when you are exposed to sunlight.

A vitamin D deficiency makes it difficult for your body to maintain sufficient levels of calcium and phosphate. When this occurs, your body produces hormones that cause calcium and phosphate to be released from your bones. When your bones lack these minerals, they become weak and soft.

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Rickets is most common in children who are between 6 and 36 months old. Children are at the highest risk of rickets because they are still growing. Children might not get enough vitamin D if they live in a region with little sunlight, follow a vegetarian diet, or don't drink milk products. In some cases, the condition is hereditary.

Rickets is rare in the United States. Rickets used to be more common, but it mostly disappeared in developed countries during the 1940s due to the introduction of fortified foods, such as cereals with added vitamin D.

2. EXPERT SYSTEM

An expert system is one of the strongest branches of Artificial Intelligence (AI) [10-14], which aims to transfer the human intelligence to computer systems through the design of software and computing devices that simulate the behavior and thinking of humans [1,3]. An expert system consists of a knowledge base and an inference engine [2, 4]. Knowledge base is generated through two main components: human expert and knowledge engineer, which convert this experience to knowledge base that consists of rules. Inference engine is an intermediary between the end user and the knowledge base, see Figure 2 for details.

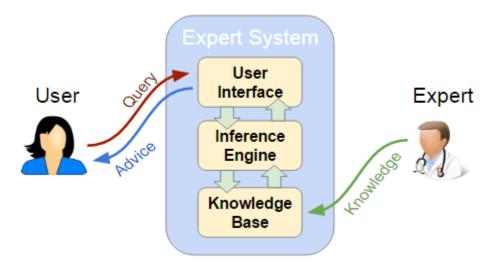


Figure 2: The figure presents the main Components of an Expert System.

The expert system for rickets diagnosis was implemented by using Simpler Level 5 Object (SL5 O) [5], which was influenced by Official Production System (OPS5) and Level 5 (L5) [5, 42]. SL5 Object is a forward chaining expert system language, multiparadigm programming language (rule based and object oriented) and pattern matching.

3. LITERATURE REVIEW

There are many clinical tasks that can be implemented by expert systems such as [12-41]:

- Help in diagnosis: When the patient's condition is complex or the person that doing the diagnosis is not an experienced one, an expert system provides meaningful diagnoses based on patient data.
- Proposing treatment: Expert system that can formulate a treatment plan based on the patient's condition and evidence-based treatment.

There are a few expert systems that diagnose rickets; Rickets Expert System (RES) architecture for early diagnosis is an expert system for diagnosing children Rickets using web based application [7], but does not include all the causes of rickets and its symptoms.

3.1 Risk Factors For Rickets

Age

Rickets is most common in children who are between 6 and 36 months old. During this time period, children usually experience rapid growth. This is when their bodies need the most calcium and phosphate to strengthen and develop their bones.

• Diet

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You have a higher risk of developing rickets if you eat a vegetarian diet that doesn't include fish, eggs, or milk. You are also at an increased risk if you have trouble digesting milk or have an allergy to the protein in dairy (lactose). Infants who are only fed breast milk can become deficient in vitamin D as well. Breast milk doesn't contain enough vitamin D to prevent rickets.

• Skin Color

Children of African, Pacific Islander, and Middle Eastern descent are at the highest risk for rickets because they have dark skin. Dark skin doesn't react as strongly to sunlight as lighter skin does, so it produces less vitamin D.

• Geographic Location

Our bodies produce more vitamin D when they are exposed to sunshine, so you are more at risk for rickets if you live in an area with little sunlight. You are also at a higher risk if you work indoors during daylight hours.

• Genes

A form of rickets can be inherited. This means that the disorder is passed down through your genes. This type of rickets, called hereditary rickets, prevents your kidneys from absorbing phosphate.

3.2 Pathophysiology

Cholecalciferol (i.e., vitamin D-3) is formed in the skin from 5-dihydrotachysterol. This steroid undergoes hydroxylation in 2 steps. The first hydroxylation occurs at position 25 in the liver, producing calcidiol (25-hydroxycholecalciferol), which circulates in the plasma as the most abundant of the vitamin D metabolites and is thought to be a good indicator of overall vitamin D status [10].

The second hydroxylation step occurs in the kidney at the 1 position, where it undergoes hydroxylation to the active metabolite calcitriol (1, 25-dihydroxycholecalciferol). This cholecalciferol, which circulates in the bloodstream in minute amounts, is not technically a vitamin but a hormone.

Calcitriol acts at 3 known sites to tightly regulate calcium metabolism: (1) it promotes absorption of calcium and phosphorus from the intestine; (2) it increases reabsorption of phosphate in the kidney; and, (3) it acts on bone to release calcium and phosphate. Calcitriol may also directly facilitate calcification. These actions result in an increase in the concentrations of calcium and phosphorus in extracellular fluid.

This increase of calcium and phosphorus in extracellular fluid, in turn, leads to the calcification of osteoid, primarily at the metaphyseal growing ends of bones but also throughout all osteoid in the skeleton. Parathyroid hormone facilitates the 1-hydroxylation step in vitamin D metabolism.

In the vitamin D deficiency state, hypocalcemia develops, which stimulates excess secretion of parathyroid hormone. In turn, renal phosphorus loss is enhanced, further reducing deposition of calcium in the bone.

Excess parathyroid hormone also produces changes in the bone similar to those occurring in hyperparathyroidism. Early in the course of rickets, the calcium concentration in the serum decreases. After the parathyroid response, the calcium concentration usually returns to the reference range, though phosphorus levels remain low. Alkaline phosphatase, which is produced by overactive osteoblast cells, leaks into the extracellular fluids, so that its concentration rises to anywhere from moderate elevation to very high levels.

Intestinal malabsorption of fat and diseases of the liver or kidney may produce the clinical and secondary biochemical picture of nutritional rickets. In such cases, disturbance in calcium homeostasis may be the consequence of renal excretion or may result from intestinal losses, as dietary calcium forms insoluble soaps with malabsorbed fats. Anticonvulsant drugs (e.g., phenobarbital, phenytoin) accelerate metabolism of calcidiol, which may lead to insufficiency and rickets, particularly in children who have darkly pigmented skin and those who are kept primarily indoors (e.g., children who are institutionalized), see Figure 3 for details.

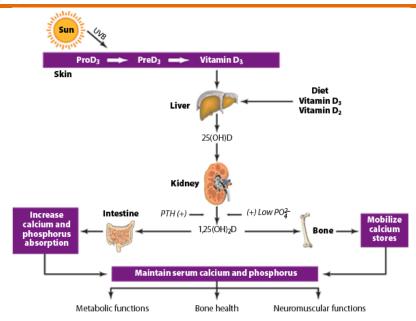


Figure 3: The figure presents Physiology of vitamin D.

3.3 TYPES OF RICKETS

- a) Vitamin D deficient rickets: there is decrease in vitamin D inside body.
- b) Vitamin D dependent rickets: there is defect in the process of vitamin D activation.
- c) Vitamin D resistant rickets: either
 - Hypophosphatemic rickets
 - End organ resistance to 1, 25 Dihydroxy Vitamin D3.

3.4 SYMPTOMS OF RICKETS

- pain or tenderness in the bones of the arms, legs, pelvis, or spine
- stunted growth and short stature
- bone fractures
- muscle cramps
- teeth deformities, such as:
 - o delayed tooth formation
 - o holes in the enamel
 - o abscesses
 - o defects in the tooth structure
 - an increased number of cavities
- skeletal deformities, including:
 - o an oddly shaped skull
 - o bowlegs, or legs that bow out
 - o bumps in the ribcage
 - o a protruding breastbone
 - o a curved spine
 - o pelvic deformities

Call your doctor right away, if your child is showing signs of rickets. When the disorder is not treated during a child's growth period, the child may end up with a very short stature as an adult. Deformities can also become permanent if the disorder goes untreated [11], see Figure 4 for details.

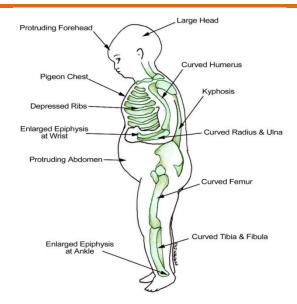


Figure 4: The figure presents Signs and symptoms of rickets.

3.5 HOW TO APPROACH TO RICKETS

In this expert system, there are three types of rickets:

- 1. Vitamin D deficient rickets
- 2. Vitamin D dependent rickets
- 3. Vitamin D resistant rickets

The present Expert system requires the user to answer the question displayed on the screen (see Figure 5 for a snapshot screen of the expert system), starting with the first question (Are patient signs and symptoms consistent with hypocalcemia?) and ending with the diagnosis of the disease and a recommendation of the treatment. This expert system not only displays to the user the diagnose of the disease but the appropriate treatment also at the end of the dialogue between the end user and the expert system. See figures 6, 7, and 8 for expert system that diagnosing the rickets [9].

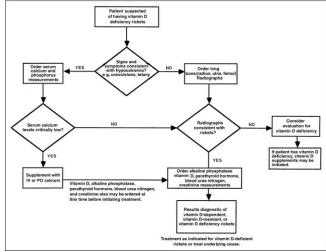


Figure 5: The figure presents the expert system for diagnosing Rickets diseases.

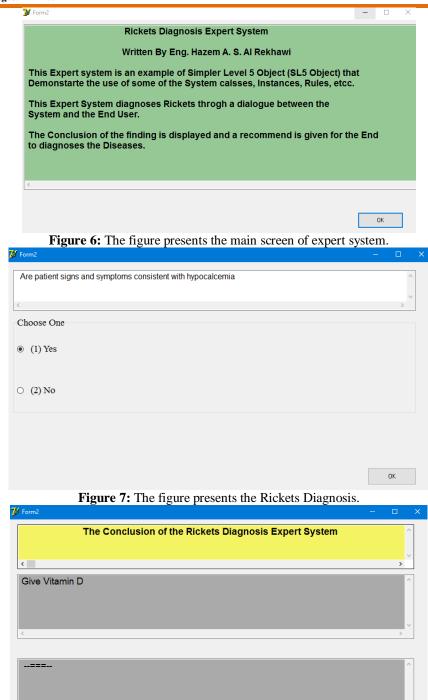


Figure 8: The figure presents the result of Rickets Diagnosis.

4. LIMITATIONS

The current expert system limited to the following rickets types: Vitamin D deficient rickets, Vitamin D dependent rickets and Vitamin D resistant rickets.

5. SYSTEM EVALUATION

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A number of doctors have tested this expert system and several patients who suffer from rickets have tested it. They were satisfied with the efficiency of the expert system. Patients with rickets who tried the expert system were marveled about the accuracy, ease and speed of the diagnosis of the rickets; because the system contains all the causes, types of rickets and easily obtained the data from the patient to diagnose his/her condition.

6. CONCLUSION AND FUTURE WORK

This paper has presented an expert system for rickets diagnosis, which provides the patients with the diagnosis, recommendation and treatment; based on the expert system knowledge base and data collected from the patients. The aim of the proposed expert system was to recognize the symptoms of the disease by the user to enable him/her to identify diseases and causes of rickets. This expert system save the patient the time and effort by allowing the patient to diagnose rickets seamlessly and easily through a simple user interface, so often is dispensed with the traditional diagnosis. This expert system was designed and developed using SL5 Object language. In fact, there is a growing in rickets rate, it became necessary to propose several expert systems to support the diagnosis of diseases and causes of rickets and recommendations and treatment. There are several directions to carry out further research, this means adding more from diseases and causes of rickets. We confined the current expert system to specializes in rickets, some of which we plan to build in the future such as expert system that combines the other causes of rickets that include Hypophosphatemic rickets, Hypocalcemic related rickets and other secondary causes of the diseases using other Expert system languages.

7. EXPERT SYSTEM SOURCE CODE

```
! Written By Hazem Awni Al Rekhawi!
ATTRIBUTE Are patient signs and symptoms consistent with hypocalcemia? COMPOUND Yes, No
ATTRIBUTE Order serum calcium and phosphate and check If the percentage is low? COMPOUND Yes, No
ATTRIBUTE Order long bone X Ray and check if Radiology consistent with Rickets? COMPOUND Yes, No
ATTRIBUTE Consider evaluation for Vitamin D Deficiency? COMPOUND Yes, No.
ATTRIBUTE start SIMPLE
INSTANCE the domain ISA domain
 WITH start := TRUE
INSTANCE the application ISA application
WITH title display := introduction
WITH conclusion display := Conc
WITH numeric precision := 8
WITH simple query text := "Is it true that:
is
 WITH numeric query text := "What is(are):
of
 WITH string query text := "What is(are):
of
 WITH time query text := "What is(are):
of
 WITH interval query text := "What is(are):
of
 WITH compound query text := "
```

WITH text :=" --===--"

```
of
 WITH multicompound query text := "What is(are):
of
INSTANCE introduction ISA display
 WITH wait := TRUE
 WITH delay changes := FALSE
 WITH items [1] := textbox 1
INSTANCE textbox 1 ISA textbox
 WITH location := 10,10,1000,350
 WITH pen color := 0.0.0
 WITH fill color := 150,200,150
 WITH justify IS left
 WITH font := "Arial"
 WITH font style IS bold
 WITH font size := 14
 WITH text :="
                        Rickets Diagnosis Expert System
                       Written By Eng. Hazem A. S. Al Rekhawi
This Expert system is an example of Simpler Level 5 Object (SL5 Object) that
Demonstarte the use of some of the System calsses, Instances, Rules, etcc.
This Expert System diagnoses Rickets through a dialogue between the
System and the End User.
The Conclusion of the finding is displayed and a recommend is given for the End User
to diagnoses the Diseases."
INSTANCE Conc ISA display
 WITH wait := TRUE
 WITH delay changes := FALSE
 WITH items [1] := title textbox
 WITH items [2] := problem textbox
 WITH items [3] := advise textbox
INSTANCE title textbox ISA textbox
 WITH location := 20,10,800,90
 WITH pen color := 0.0.0
 WITH fill color := 243,243,100
 WITH justify IS center
 WITH font := "Arial"
 WITH font style IS bold
 WITH font size := 14
 WITH text := "The Conclusion of the Rickets Diagnosis Expert System"
INSTANCE problem textbox ISA textbox
 WITH location := 20,110,800,130
 WITH pen color := 0,0,0
 WITH fill color := 170,170,170
 WITH justify IS left
 WITH font := "Arial"
 WITH font size := 14
```

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INSTANCE advise textbox ISA textbox

WITH location := 20,280,800,130

WITH pen color := 0,0,0

WITH fill color := 170,170,170

WITH justify IS left

WITH font := "Arial"

WITH font size := 14

WITH text :=" --===--"

RULE RO

IF start

THEN ASK Are patient signs and symptoms consistent with hypocalcemia?

RULE R1

IF Are patient signs and symptoms consistent with hypocalcemia? IS Yes

THEN ASK Order serum calcium and phosphate and check If the percentage is low?

RULE R1a

IF Are patient signs and symptoms consistent with hypocalcemia? IS No

THEN ASK Order long bone X Ray and check if Radiology consistent with Rickets?

RULE R2

IF Order long bone X Ray and check if Radiology consistent with Rickets? IS Yes

THEN text OF problem textbox := "Order alkaline phosphatase vitamin D"

RULE R2a

IF Order long bone X Ray and check if Radiology consistent with Rickets? IS No

THEN ASK Consider evaluation for Vitamin D Deficiency?

RULE R3

IF Consider evaluation for Vitamin D Deficiency? IS Yes

THEN text OF problem textbox := "Give Vitamin D"

RULE R3a

IF Consider evaluation for Vitamin D Deficiency? IS No

THEN text OF problem textbox := "Search for other Causes"

RULE R4

IF Order serum calcium and phosphate and check If the percentage is low? IS Yes

THEN text OF problem textbox := "Supplement Calcium"

RULE R4a

IF Order serum calcium and phosphate and check If the percentage is low? IS No

THEN ASK Order long bone X Ray and check if Radiology consistent with Rickets?

END

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