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Unraveling the Rare Connection: Rickets Syndrome and Secondary Osteoarthritis - An Intriguing Case Study

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ABSTRACT

Osteoarthritis (OA) is a degenerative joint disease characterized by progressive destruction and loss of articular cartilage. Osteoarthritis of the knee can be divided into two types, namely primary and secondary. Primary osteoarthritis is articular degeneration without an obvious cause, while secondary osteoarthritis is a consequence of abnormal force concentration in the joint due to post-traumatic causes or abnormal articular cartilage, such as rheumatoid arthritis (RA). Rickets syndrome (rickets) is characterized by mineralization defects and widening of the epiphyseal plate. The mineralization defect that occurs in patients with rickets certainly affects bone and cartilage formation. These bone formation abnormalities can inhibit movement and cause pain. A patient came from the orthopedic clinic for consultation regarding knee pain since 6 months ago. This complaint arose starting with the patient fell on his right-side knee. Previously, the patient could walk, but because the patient had fallen and hit his right knee, the patient immobilized his knee for 3 months. After immobilization, the patient's leg felt stiff, which made the patient unable to walk again. The patient previously had a rickets syndrome (rickets) disorder diagnosed by an orthopedic doctor since the patient was 6 years old. Patients with rickets syndrome have a higher risk of secondary osteoarthritis. This is due to mineralization defects that affect bone and cartilage growth. These growth abnormalities will affect bone strength and cartilage growth, so that even with minimal fractures can cause severe osteoarthritis in patients with rickets disorders.

Keywords: ricket syndrome; osteoatritis; mineralization defect; human and medicine.

INTRODUCTION

Osteoarthritis (OA), also known as degenerative joint disease, is usually caused by progressive deterioration and loss of articular cartilage. Osteoarthritis of the knee can be divided into two types, namely primary and secondary. Primary osteoarthritis is articular degeneration without an obvious cause, while secondary osteoarthritis is a consequence of abnormal force concentration in the joint due to post-traumatic causes or abnormal articular cartilage, such as rheumatoid arthritis (RA). (Xia et al., 2014; Glyn-Jones et al., 2015; Hussain et al., 2016)

Rickets is a disease that can be inherited or acquired. Vitamin D, calcium, and phosphorus are the main factors that influence bone maturation and mineralization. Mineralization defects can lead to rickets and/or osteomalacia. Rickets is characterized by mineralization defects and widening of the epiphyseal plate, while osteomalacia is a defect in the mineralization of the bone matrix. Both Rickets and osteomalacia usually occur together in children. Rickets occurs exclusively in children, while adults develop osteomalacia after dilation of the epiphyseal plate. (Genetics, 2020; Haffner et al., 2022)

Mineralization defects that occur in patients with rickets certainly affect bone and cartilage formation. Such bone formation abnormalities can hinder movement and cause pain. These bone abnormalities become more pronounced, especially if the patient engages in activities that use the knee for support. If left untreated, these abnormalities tend to worsen over time. (Creo et al., 2017; Lambert and Linglart, 2018)

This case report discusses a rickets patient who developed secondary osteoarthritis due to trauma to the patient's knee. This case is interesting to discuss because the bone demineralization and bone deformity that occur will be associated with damage to the joint bone and cartilage of the patient's knee, making it easier for osteoarthritis to occur.

CASE REPORT

A patient came from the orthopedic clinic for consultation regarding knee pain since 6 months ago.

This complaint arose starting with the patient fell on his right-side knee. Previously, the patient could walk, but because the patient had fallen and hit his right knee, the patient immobilized his knee for 3 months. After immobilization, the patient's leg felt stiff, which made the patient unable to walk again. The patient previously had a rickets disorder diagnosed by an orthopaedic doctor since the patient was 6 years old.

It was known that the patient's younger brother had a similar disorder. The patient had consulted an orthopaedic doctor to overcome his illness, but the patient was only given treatment in the form of vitamins and calcium. The patient had previously experienced pain in the hip bone and complained that it was difficult to walk. After consultation, the orthopaedic doctor advised the patient to perform surgery on the hip bone and was advised to perform surgery. However, due to the cost of the patient had to wait until finally the patient fell and causing the patient's knee to hurt, but after the patient consulted again related to his knee the patient was advised to repair the hip bone first and perform hip bone replacement surgery. After 3 months later, the patient returned for control and continued surgery on his knee. From the physical examination, it was found that the patient looked mildly ill. Blood pressure 120/80 mmHg, pulse 81 ×/min, respiratory frequency 21 x/min, temperature 36.7 OC, BMI 21.8 kg/m2. Eyes, ears, nose, and mouth within normal limits. The neck has no enlarged lymph nodes.

Right and left vesicular lung sounds. Heart sounds on auscultation examination are regular. Abdominal auscultation of bowel noise (+) within normal limits. Neurological status was found to be normal physiological reflexes and negative pathological reflexes. Local status in the thoracic region was found to appear as breastbone projection, genue dextra et sinistra appeared valgum, knock knee obtained the same skin color as the surrounding, no mass, edema, or cyanosis was found. Palpation examination found no lumps or swelling, crepitation (+/+), tenderness (+/+). The patient's genue range of movement is limited to 0-5 degrees.

This patient underwent a supporting examination in the form of laboratory tests and radiological examinations in the form of x-rays of the AP / Lateral femur regio and AP genue regio dextra et sinistra. Laboratory support examination was obtained.

FIGURE 1: Patient Clinical Appearance.

This patient underwent a supporting examination in the form of laboratory tests and radiological examinations in the form of x-rays of the AP / Lateral femur regio and AP genue regio dextra et sinistra. Laboratory support examination was obtained.

Table 1: Laboratory Examination of Patient 27/09/22.

Assesment	Result	Refferal	Unit
Hematolog			
Hemoglobin	15,1	14-17	g/dL
Hematocrit	45	45-55	%
Erythrocytes	5,4	4,7-6,1	10^6 /mm^3
Leukocytes	6,4	4,5-10,5	10^3 /mm^3
Platelets	279	150-450	10^3 /mm^3

Assesment	Result	Refferal	Unit
Hematolog			
MCV	83	80-100	fL
MCH	28	27-31	pg
MCHC	34	32-36	%
Diff Count			
Eosinophils	8	0-6	%
Basophils	0	0-2	%
Rod Netrophils	0	02-Jun	%
Segment Netrophils	65	50-70	%
Lymphocyte	22	20-40	%
Monocyte	5	02-Aug	%
Hemostasis			
PT	14	11,50 – 15,50	Detik
APTT	30,9	26,00 – 37,00	Detik
Imunoserology			
HbsAG	Non-Reactive	Non-Reactive	
Clinical Chemistry			
Randome Blood Glucose Level	125	<200	Mg/dl
Ureum	30	13-43	Mg/dl
Chloride	0,65	0,67-1,17	Mg/dl
Electrolytes			
Sodium	144	132-146	mmol/l
Potasium	3,9	3,7-5,4	mmol/l
Chloride	108	98-106	mmol/l
Calcium	7,7	8,6-10,3	Mg/dl

Laboratory examination of the patient's blood found that the patient had hypocalcemia. Subsequently, an X-ray examination of the os femur was performed on this patient.

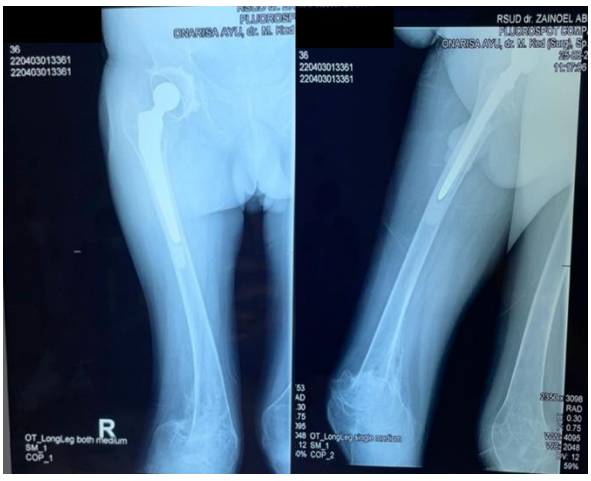

FIGURE 2: X-ray Femur AP/Lateral (Post-ORIF).

X-ray examination of the dextra femur AP/lateral obtained total hip replacement using acetabulum and cemented femoral stem with good position. There was a transverse complete fracture at the distal 1/3 of the dextra femoral os displacement towards the anterolateral.

FIGURE 3: X-ray Genu AP/Lateral (Pre-Operative).

On x-ray examination of the dextra femur AP/lateral postoperative ORIF os the Femur. Obtained implants in the dextra hip joint and plating in the dextra distal femur with good position, accompanied by narrowing with a picture of kissing lesion in the dextra genu os. After taking a history, physical examination, and supporting examination, the patient was diagnosed with secondary osteoarthritis.

FIGURE 4: X-Ray Genu AP/Lateral Post-Operative.

The patient was treated for 3 months to wait for the results of recovery after ORIF os the femur surgery. Furthermore, the patient did a re-control to reassess the treatment results. Furthermore, an X-ray examination of the AP / Lateral femur was carried out.

FIGURE 5: X-Ray Genu AP/Lateral Pasca ORIF (After 3 months observation).

X-ray examination of the dextra Ap/lateral femur after ORIF os the Femur surgery after 3 months of observation. A callus (+) picture was found with normal bone trabeculation, with a gap, and the joint surface looked good, and there was no soft tissue mass/swelling.

After 3 months of observation, a total knee replacement with a long stem was performed to correct the patient's secondary OA. The following are the postoperative X-rays of the patient's total knee replacement.

FIGURE 6: X-Ray Genu AP/Lateral After Total Knee Replacement.

The follow-up X-ray examination after total knee replacement surgery revealed the attachment of a long stem knee prosthesis to the distal os of the femur and the proximal os of the tibia dextra.

DISCUSSION

a. Correlation between Osteopenia and Abnormal Gait.

The patient came with complaints of pain and stiffness in the patient's knee joint, which was preceded by a previous trauma. Complaints of joint stiffness accompanied by pain in the patient's knee made it difficult for the patient to walk and stand, thus reducing the patient's quality of life.

Osteopenia that occurs in patients causes a decrease in density rather than bone density, which causes with a little pressure force on the bone, makes the bone easy to fracture. Decreased calcium levels in the body cause reduced ATP production, which affects the lack of energy that can be used to strengthen muscles, so that it can weaken the proximal muscles, which will affect the patient's walking.

b. Pathophysiology of Ricket Syndrome.

The patient was diagnosed with Rickets 1 year ago. It is known based on anamnesis that the patient has suffered from abnormalities since the patient was 6 years old. This assessment is seen from the difference in posture, accompanied by a clinical appearance that is different from his peers.

The patient's own diagnosis is based on the bone growth and clinical appearance of the patient. According to the theory, rickets is a disease caused by mineralization defects rather than bone, characterized by disruption of growth activity in the epiphyseal plate. This will certainly relate to the formation of bone and cartilage itself.

Patients with rickets will have a characteristic appearance in the form of a chest that resembles a breastbone projection, thickening of the bones of the wrists and feet, accompanied by a valgum or knock knee appearance. (Bitzan and Goodyer, 2019)

Local status in the thoracic region was found to appear as breastbone projection, genue dextra et sinistra appeared valgum, knock knee obtained the same skin color as the surrounding, no mass, edema, or cyanosis was found. Palpation examination found no lumps or swelling, crepitus (+/+), tenderness (+/+). The patient's genue range of movement is limited to 0-5 degrees. Osteopenia that occurs in the bones of patients with rickets will affect the growth that occurs in the epiphyseal plate. Patients with rickets will have abnormalities in the patient's bone cartilage. The cartilage in the epiphyseal plate cannot be ossified. The non-oscillated epiphyseal plate causes incomplete formation of the epiphyseal plate growth, so that it interferes with bone growth, which causes the patient to be of short stature. The weight-bearing bone support with the osteopenia condition of the rickety patient will cause the bowed legs condition in the patient. (Creo et al., 2017; Lambert and Linglart, 2018; Genetics, 2020)

Supporting examination in the form of a blood lab examination found that the patient had hypocalcemia with a calcium level in the patient's blood of 7.7 mg/dl. However, the patient was not tested for vitamin D levels and blood phosphatase levels. According to the theory, because rickets is a disease due to mineralization defects in the bone, of course, from the blood laboratory examination of patients with rickets have deficiencies in calcium levels, phosphatase, and also levels of vitamin D. (Lambert and Linglart, 2018; Genetics, 2020)

Patients with rickets in developing countries come from malnutrition that occurs in patients. Nutrient deficiency in the form of vitamin D will affect the absorption of phosphate and calcium levels in the bones, the lack of vitamin D levels stimulates parathyroid hormone to continue to release calcium levels into the bloodstream, causing osteopenia. (Creo et al., 2017)

A supporting examination in the form of a genu x-ray was performed on the patient to assess abnormalities for complaints of pain and stiffness in the knee. This was confirmed by the picture of narrowing of the joint gap, accompanied by osteophytes and also fractures seen on x-ray examination of the genu. (Creo et al., 2017; Haffner et al., 2022; Trombetti et al., 2022)

C. Influence of rickets on the occurrence of secondary osteoarthritis.

According to the theory, secondary osteoarthritis can occur due to various causes, including trauma and abnormalities in joint cartilage formation. In patients with rickets, there is certainly a disturbance in bone density and cartilage formation, which is related to the osteoarthritis condition that occurs in patients.

The patient's secondary osteoatritis condition was caused by minimal trauma followed by fracture due to osteopenia caused by demineralization of the bone and abnormalities in the formation of the patient's joint cartilage. (Xia et al., 2014; Abramoff and Caldera, 2020)

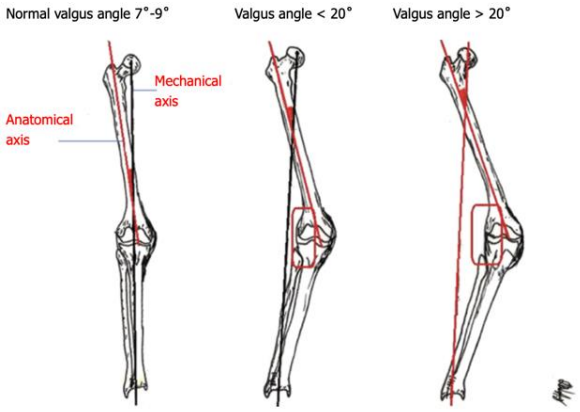

FIGURE 7: Comparison between Normal Valgus Angle and Abnormal Valgus Angle.

The knock knee or valous condition that occurs in the patient results in a significant buildup in the weight-bearing joint, continuing to occur in the patient's knee, causing disruption of the tibiofemoral joint of more than 20%. It is also seen that the patient has narrowed the lateral and medial sides of the tibiotemoral joint until tibial glide occurs in the joint, which makes the patient feel limited in performing active and passive flexion movements, so that operative action is needed. (Mandl, 2019)

Although the patient already had grade 4 genu A with the appearance of a kissing lesion in the genu joint, because this patient had fractured the distal part of the femoral os, ORIF was performed in this patient first to restore the mechanical axis in the patient before total knee replacement was bertormed to repair the condition of the femoral os that had fractured.

According to the theory of grade 1-3 osteoatritis. Non-operative therapy can be carried out by administering NSAID drugs or using orthoses, physical therapy, and lifestyle changes. However, cases with grade 4 osteoatritis require definitive therapy in the form of total knee replacement surgery to reduce the destruction that occurs due to cartilage friction, especially in the knee joint. (Xia et al., 2014; Hussain et al., 2016; Abramoff and Caldera, 2020; Trombetti et al., 2022)

D. Comparison Before and After Total Knee Replacement

This patient has undergone an operative procedure in the form of a total knee replacement with a long stem, which aims to strengthen the prosthesis on bones that have a history of fracture and osteopenia. This action has been completed, and of course, the quality assessment of the actions taken can improve pain factors, function, and mobility, range of movement, along with radiographic evaluation.

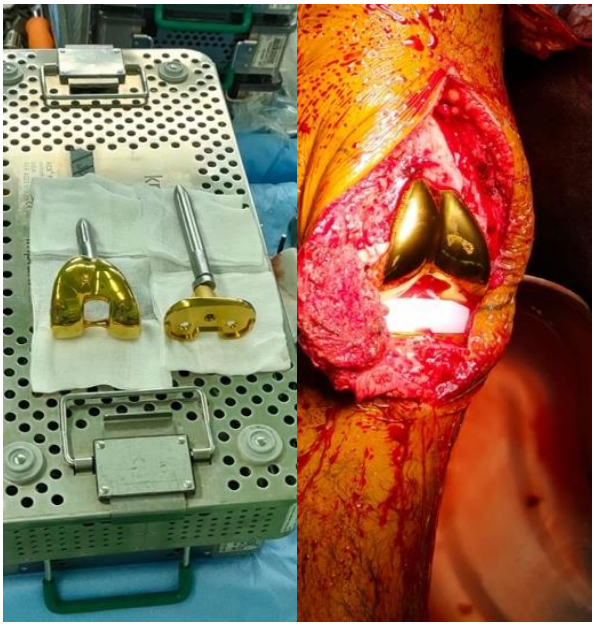

FIGURE 8: Knee Long-stem Prostheses.

- ***Pain Levels***

Significant changes occurred after surgery on the patient; it is known that the patient's pain has been significantly reduced after surgery. This comparison can be seen from the patient's condition after observation for 1 month postoperatively. The patient underwent active and passive pain provocation found the patient did not complain of pain, decreased visual analog scale from 4 to 0.

- ***Function and Mobility***

Knee osteoarthritis that occurs in patients causes interference for patients to walk or do activities in daily life. After surgery, physiotherapy, and observation for 6 months, the patient was evaluated in terms of walking ability. This assessment was carried out by walking, preceded by a cane aid using 2 sticks, I stick, and without using aids. From the assessment, it was found that the patient had improved quality and function in walking.

FIGURE 9: Patient Walk Quality Improvement.

It can be seen from the picture above that there is a significant improvement in changes in the quality of the patient's walking during physiotherapy and regular observation for 1 month. After 6 months, the patient can walk and move again, and also work.

- ***Range of Movement***

Clinical examination of the patient has also shown a change in the range of motion in the patient's knee. It was found that the patient was able to perform maximum flexion and extension after physiotherapy.

- ***Radiographic Evaluation***

FIGURE 10: X-Ray Before and After Total Knee Replacement.

X-ray radiology examination of the genu obtained improvement, marked by widening the joint gap in the genu os joint that has been installed with a long stem knee prosthesis in a good position, and there is an improvement in the mechanical axis that is significant enough to reduce the valgus angle abnormality in the patient.

Strengths and limitations

The case study presents a comprehensive and detailed account of the progression of secondary osteoarthritis in a patient with rickets syndrome. The unique correlation between the two conditions is a rare finding, allowing for a clear cause-and-effect analysis. The report highlights the surgical and rehabilitation interventions undertaken to treat the patient, showcasing step-by-step progress throughout the treatment process. The successful outcome of the surgery and rehabilitation is evident in the significant improvement in the patient's quality of life.

CONCLUSIONS

Patients with rickets have a higher risk of secondary osteoarthritis. This is due to mineralization defects affecting both bone and cartilage growth. A consequence of abnormal force concentration at the joint due to trauma or abnormal articular cartilage. Minima fracture facilitation, accompanied by valgus-knee deformity in rickets patients, leads to abnormal support at the os tibiofemoral joint, thus increasing the risk of osteoarthritis.

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AUTHOR CONTRIBUTION

MDD and AINA were instrumental in the design of this study, including the preparation and drafting of the manuscript. They actively wrote the manuscript and were involved in its thorough revision. Moreover, they provided supervision throughout the entire research process, ensuring the study's quality and accuracy. Their significant contributions played a crucial role in bringing this case study to fruition.

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