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# Imaging of nail-patella syndrome

## 以影像描述甲髕綜合徵

Nail-patella syndrome is a rare disorder, which is inherited as an autosomal dominant trait. This condition is also known as hereditary osteo-onychodysplasia or Fong's syndrome. Posterior iliac horns are commonly found in this syndrome and are considered pathognomonic. In this report, we describe the appearance, location, and structure of iliac horns with respect to radiography and magnetic resonance imaging.

甲髕綜合徵是一種非常罕見的紊亂病症，由常染色體顯性遺傳造成，又稱為遺傳性爪甲骨發育不全或方氏綜合徵。常見的病理特徵是後髕骨突出。本報告利用放射學和磁共振成像兩種途徑，描述髕骨突出的形成、發病位置以及結構。

### Introduction

Nail-patella syndrome (NPS), also known as hereditary osteo-onychodysplasia or Fong's syndrome, is a hereditary disorder with an autosomal dominant trait. This condition is caused by mutations in the *LMX1B* gene,<sup>1</sup> and has a very low incidence of about 4.5 per million in the United States.<sup>2</sup> The condition affects the nails, skeletal system, kidneys, and eyes.<sup>3</sup> The skeletal features include absent or hypoplastic patellae, patellar dislocations, elbow abnormalities, talipes, and iliac horns as seen on radiographs. These iliac horns are considered to be pathognomonic feature of NPS. In an autopsy study by Darlington and Hawkins,<sup>4</sup> the localisation of the iliac horns in NPS was determined to be at the site of attachment of the gluteus medius muscles, but this finding was not proved by the in-vivo imaging. The magnetic resonance imaging (MRI) of the features of this syndrome has been mentioned in only one report<sup>5</sup>; however, no images were actually presented. Therefore, we present the findings of abdominopelvic MRI of an NPS case to determine the exact relationship between gluteus muscles and iliac horns, and to reveal any additional morphological kidney abnormalities.<sup>6</sup>

#### Key words:

Magnetic resonance imaging;  
Nail-patella syndrome;  
Radiography

#### 關鍵詞：

磁共振成像；  
甲髕綜合徵；  
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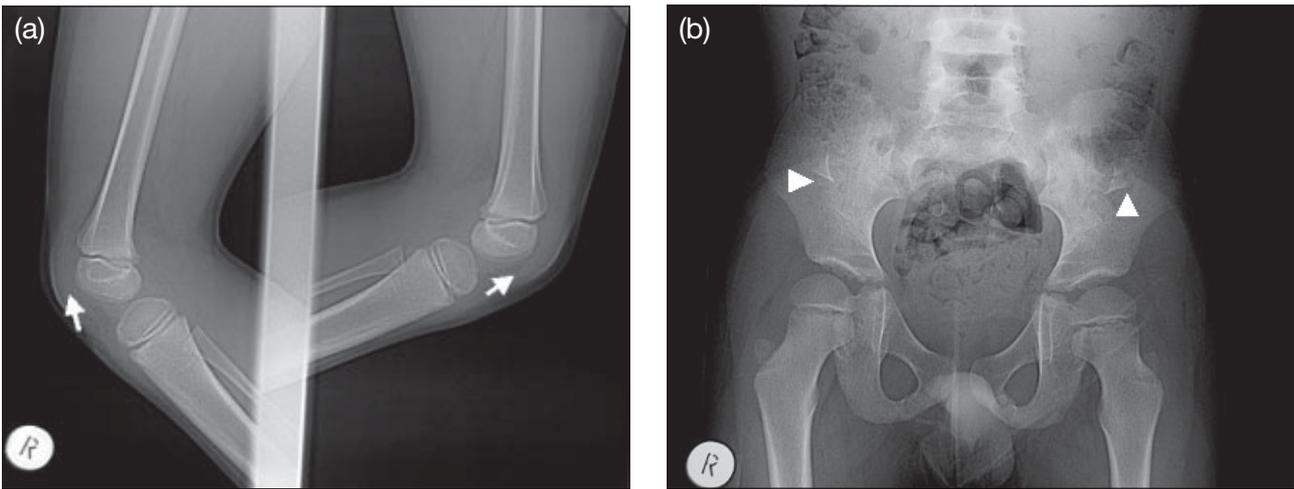
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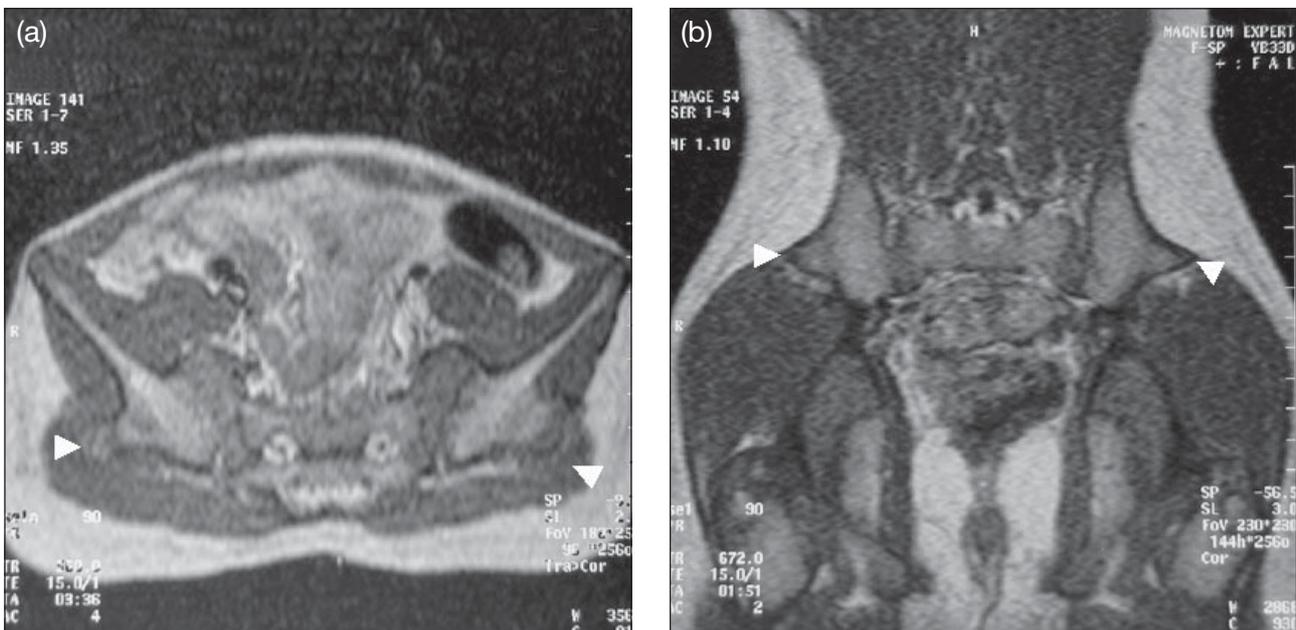
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### Case report

A 7-year-old boy presented with nail dysplasia, which is the characteristic external finding of NPS. His sister, mother, and grandfather were also known to have had similar symptoms. His physical examination, mental status evaluation, and routine laboratory analyses were unremarkable with the exception of the external finding as mentioned above. Radiography of the hand showed delayed bone ageing, and an elbow radiograph revealed hypoplasia of the radial epiphysis. The mediolateral radiograph revealed that both patellae were absent (Fig 1a), and pelvic radiography confirmed iliac horns with accompanying skeletal features. These features consisted of elongated femoral necks and bilateral coxa valga deformity (Fig 1b). An abdominopelvic MRI was conducted to reveal any related abnormalities of other organs (specifically the kidneys<sup>6</sup>) and to observe musculoskeletal associations of the iliac horns that were not



**Fig 1. (a) Mediolateral views of the knees showing the absence of both patellae; and (b) anteroposterior view of the pelvic radiograph showing bilateral iliac horns, elongated femoral necks, and bilateral coxa valga**



**Fig 2. (a) Axial and (b) coronal magnetic resonance images showing iliac horns located at the site of attachment of the gluteus medius muscles and their continuity with the iliac bones**

yet presented in vivo. To avoid unnecessary ionising radiation, computed tomographic and intravenous pyelographic imaging were not performed. Magnetic resonance imaging clearly revealed cortical and medullar components of the iliac horns, and their continuity with the corresponding iliac bones structures (Fig 2). The iliac horns were shown to be located at the insertions of gluteus muscles; however, the kidneys were morphologically normal. Considering the pathognomonic finding of iliac horns and the presence of the associated features, the patient was diagnosed with NPS.

## Discussion

Iliac horns, whose accessory outgrowths extend backwards and outwards from the mid-part of the iliac bones, are usually bilateral<sup>2,5,6</sup> and localised as the site of attachment of the gluteus medius muscles.<sup>4</sup> The horns consist of cortex and medulla that gradually blend with the normal iliac cortex. This continuity explains why these lesions do not appear rounded on anteroposterior plain films, because the X-ray beam is not at a tangent to the tissue interface medially, but the lesions can be seen clearly with the MRI technique

(Fig 2). These distinct characteristic lesions are unique to humans and occur in approximately 80% of cases of NPS, but are observed only in this condition.<sup>7</sup> Nail-patella syndrome is sometimes appropriately referred to as iliac horn syndrome.<sup>5</sup> Other than proving conclusively that the diagnosis of this syndrome is correct, iliac horns have no effect on gait and they do not need to be treated.<sup>8,9</sup>

In this case, the presence of iliac horns was shown in vivo using MRI. Patients with NPS have a variety of additional skeletal abnormalities including patellar hypoplasia or patellar dislocation (Fig 1a), delayed bone ageing, and elbow deformities. The hypoplasia of the radial head or the capitellum (in some cases both) causes subluxation or dislocation, and an inability to fully extend, pronate, or supinate the forearms.<sup>5,10,11</sup> In our patient, most of these previously reported abnormalities were also present.

Many patients with NPS seek medical attention for seemingly unrelated pathologies. In many instances, the radiologist may be the first to make the clinical diagnosis based on the findings from the imaging examinations. Moreover, early diagnosis is of particular importance in infants, because nail and patellar abnormalities may be overlooked during the first few years of life and may not be obvious until childhood. If the diagnosis of NPS is indicated, the presence of the iliac horns can confirm this syndrome.

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## Corrigendum

"Screening of dementia in Chinese elderly adults by the clock drawing test and the time and change test" (February 2005;11:13-9). We have been informed by the corresponding author of this paper that the qualifications of Dr Chan should have been the following:

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