Case Report

www.ijrhs.com
ISSN (o):2321–7251

Nail patella syndrome – a case report

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Submission Date: 20-04-2014, Acceptance Date: 25-04-2014, Publication Date: 30-04-2014

How to cite this article:

Vancouver/ICMJE Style

Roy S. Nail patella syndrome – a case report. Int J Res Health Sci [Internet]. 2014 Apr 30;2(2):678-82. Available from http://www.ijrhs.com/issues.php?val=Volume2&iss=Issue2

Harvard style

Roy, S. Nail patella syndrome – a case report. *Int J Res Health Sci*. [Online] 2(2). p.678-82 Available from: http://www.ijrhs.com/issues.php?val=Volume2&iss=Issue2

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Abstract

Nail patella syndrome or hereditary osteo-onycho-dysplasia is an autosomal dominant disorder characterized by dystrophic nails, hypoplastic or absent patellae, hypoplastic elbows and presence of pathognomonic iliac horns with or without renal involvement.The incidence is 22 per million inhabitants live birth children. Although most of the series mention posterior dislocation of the radial head but none mention its management, but we manage it as per patients demand and the case is being reported.

Key words: Nail patella syndrome; uncommon; management

Introduction

Nail patella syndrome or hereditary osteo-onycho-dysplasia is an autosomal dominant disorder characterized by dystrophic nails, hypoplastic or absent patellae, hypoplastic elbows and presence of pathognomonic iliac horns with or without renal involvement. Hereditary nature of the condition was first described by Sedgwick (quoted by Little in 1897).Turner described the syndrome in detail in 1933. Autosomal dominancy with 100% penetrance of this syndrome is linked to aberrancy on human chromosome 9's q arm 9q34. The disorder is linked to the ABO blood group locus.The incidence is 22 per million inhabitants live birth children. Although most of the series mention posterior dislocation of the radial head but none mention its management, but we manage it as per patients demand and the case is being reported.

Case report

A 28 yrs female presented in our OPD in March 2010 with pain and restriction of movement of right elbow for last 1 yr. There was no h/o trauma or massage to the right elbow. Family h/o nail changes. On examination we found short stature of the patient, hypoplastic and atrophic finger nails,

posterior dislocation of right radial head, with range of movement of right elbow -30 to 90 degree of flexion, & forearm supination 0 to 40 degree, pronation 0 to 30 degree, hyperextension of PIP joints of fingers, both the patella are small, high up and dislocates with each and every 300 flexion of knee (Habitual dislocation), and abnormal bony prominence was palpable on the posterolateral aspect of both ilium. X-ray of right elbow shows posteriorly dislocated dystrophic radial head. X-ray of both knee shows small, high up patellae and dislocated position(in skyline view).X-ray of pelvis shows presence of bilateral iliac horns on the posterolateral aspect of both ilium. Blood examination shows normal urea, creatinine and total protein. Urine examination shows mild proteinuria and presence of red cell cast (microscopic haematuria). Ultrasonography of KUB region was normal. We diagnosed the case as posterior dislocation of right elbow in a case of nail patella syndrome. After proper counseling of the patient, she demands treatment of the dislocated radial head. So we treat the patient with right radial head excision through Kocher‘s approach. POP back slab was given for 7 days followed by sling upto 3 weeks postoperatively and then active elbow exercises started. Regular follow up was done at 3 months interval. At two and half years follow up patient had no elbow, wrist or knee pain. Right elbow flexion 20 degree to 130 degree, forearm supination and pronation are full.

In the family history patients grandmother had hypoplastic fingernails and died at the age of 45 yrs due to kidney disease. Patient has two children both are girl and younger one (7yrs) has hypoplastic nails.

Discussion

Nail patella syndrome was first reported from India by Maini and Mittal (1966) [1]. Palacios proposed the term hereditary osteo-onycho-arthro-dysplasia [2]. Clinical tetrad dysplasia of thumbnail, elbow, patellae and iliac horns are main signs [3,4]. Posterior dislocation of radial head, thickening of axillary border of scapula and minor deformities of wrist are the other common findings reported [5].Bilateral congenital dislocation of hip in absence of iliac horns are also reported [6]. Flexion deformity of hips and hyperextension of interphalegeal joints are also noted [7]. Maini found flexion contracture of distal interphalegeal joints in 7 cases. Scoliosis was also noted in some cases [3]. Renal affection ranging from asymptomatic proteinuria, recurrent urinary tract infection, nephrotic syndrome, chronic glomerulonephritis, hypertension to end stage renal disease have been reported [8,9].

Autosomal dominancy with 100% penetrance of this syndrome is linked to aberrancy on human chromosome 9's q arm 9q34. The disorder is linked to the ABO blood group locus[10].Both sexes are equally affected and no generation can skip the disease[1,5].In previously reported cases nail abnormalities are present in more than 90% of cases and nails may be absent, hypoplastic or dysplastic, and toe nails may be normal. Knee and elbow abnormalities are present in 90% of cases. Patellae may be absent or hypoplastic and may be dislocated in 30% of cases. Complications such as arthritis and knee effusion can cause knee pain. Common elbow symptoms are attributed to radial heads that are typically hypoplastic leading to dislocation. The distal ends of humerus are also hypoplastic and posterior processes limits extension, supination and pronation. Iliac horns are observed in 30% to 70% cases and are pathognomonic. Other bone anomalies affecting the ankle, feet, wrist, scapula, skull and spine has been described. Renal symptoms are present in approximately 50% of cases. Most frequent symptoms are proteinuria, haematuria, nephrotic syndrome and hypertension. End stage renal disease develops in approximately 30% of patients.

Conclusion

This type of uncommon syndrome may require judicious management depending on patients demand, as in this case patient demands for her dislocated radial head but not for the habitual dislocation of patellae.

Clinical message: The uncommon Nail patella syndrome may be treated as per patients demand..

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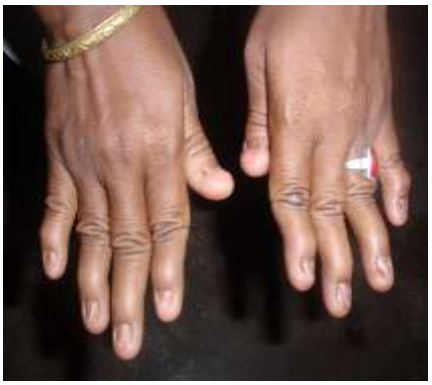
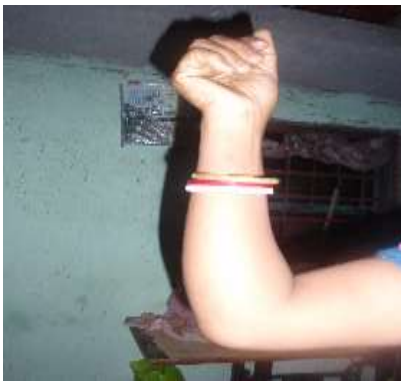
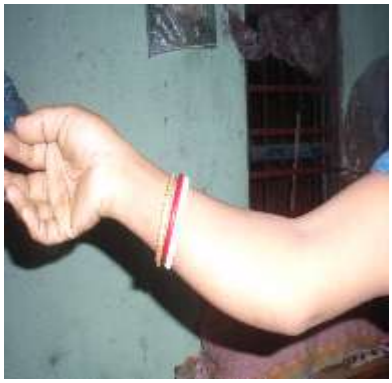
Figures:



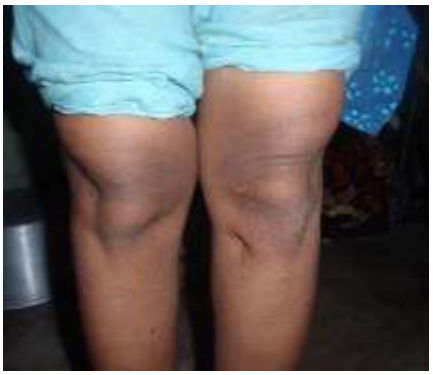
Hypoplastic Nails



Elbow Flexion



Elbow extension



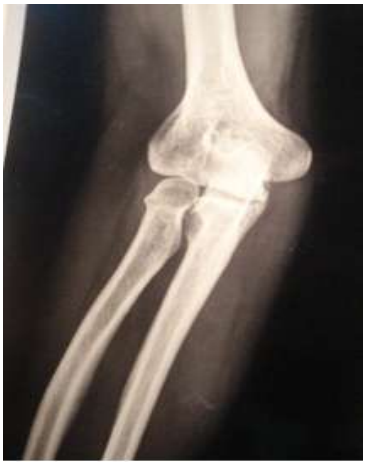
Patella

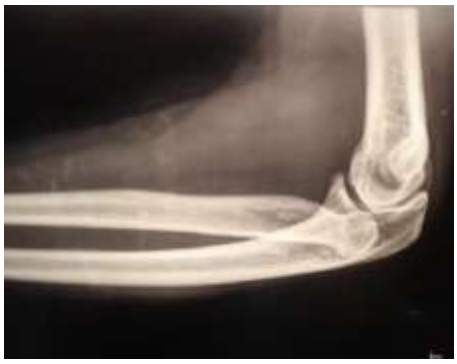
Acknowledgment

Authors acknowledge the immense help received from the scholars whose articles are cited and included in references of this manuscript. The authors are also grateful to authors/editors/publishers of all those articles, journals and books from where the literature for this article has been reviewed and discussed.

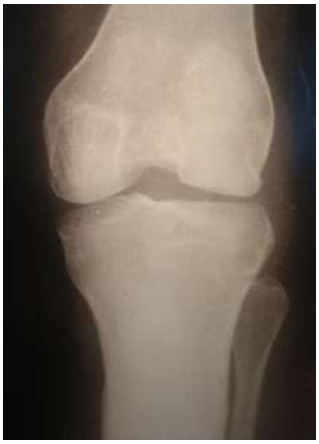
Competing interest: The authors declare that they have no competing interests

Source of Funding: Nil

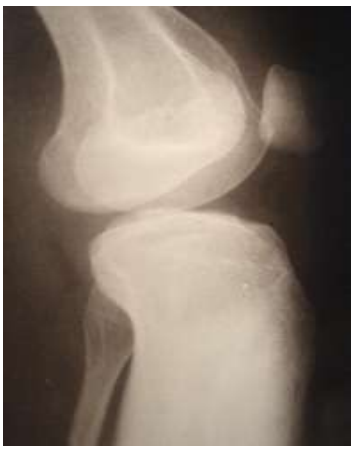


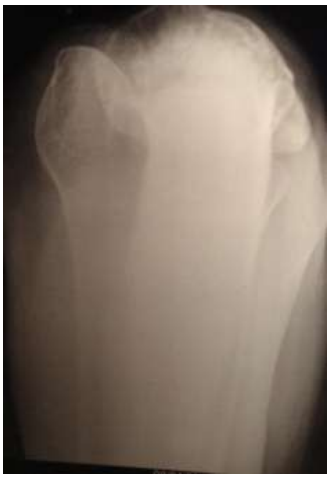


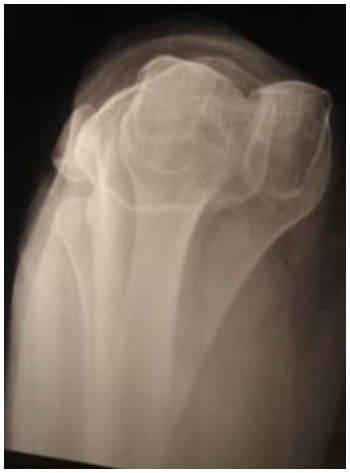
X-ray of Right Elbow



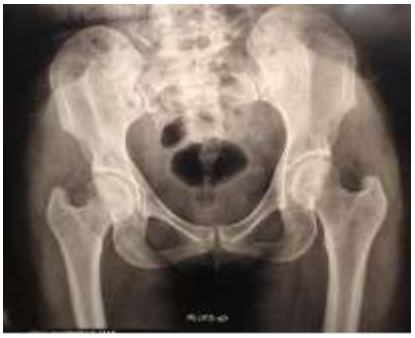
X-ray of Both Knee

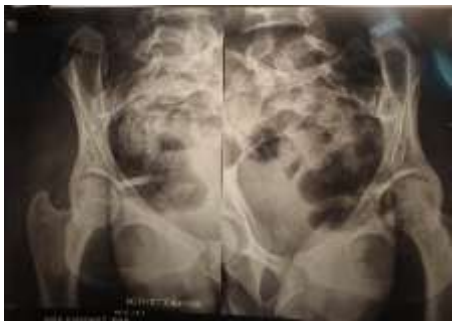




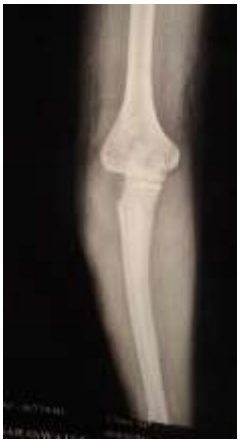


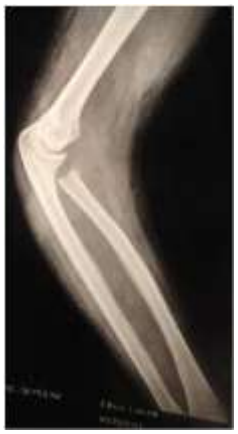
X-ray of Pelvis





POST OP X-RAY





Two and half year follow up