FAMILIAL CASES OF NAIL-PATELLA SYNDROME (NPS)

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Introduction

✓ Nail Patella Syndrome (NPS) is an hereditary osteo-onycodysplasia.
✓ (NPS) involves a classic tetrad of changes in the nails (the most constant feature), knees, elbows, and the presence of iliac horns (iliac exostosis).

✓ Frequency
✓ estimated of 1/45,000 live births
✓ NPS has been reported in patient worldwide.

✓ Etiology
✓ NPS is determined by an autosomal dominant mutation (AD) in LMX1B gene (9q34.1).

✓ The major clinical features:
✓ nail dysplasia with triangular lunula, absent/small nails of fingers and toes
✓ hypoplastic or absent patellas
✓ dysplastic elbows (with increased carrying angle of elbow)
✓ iliac exostosis (iliac horns)
✓ hyperextensible joints
✓ abnormal gait
✓ pelvis anomaly
✓ spinal and chest wall problems

✓ Other clinical features
✓ ocular: glaucoma, ocular hypertension
✓ nephropathy
✓ gastrointestinal involvement:
✓ neurological problems
✓ vasomotor problems:
✓ dental anomalies

Method

✓ We present a family with NPS
✓ The proband was B.M., female, 50 years old (fig. 1).
✓ Familial history revealed many other cases (7 cases) with the same clinical features, with AD inheritance: the father of the proband, two brothers of her (unexamined) and their children (fig. 2)

✓ We also examined the proband nephews (B.G. and B.C., the children of her brother B.I.) who present the features of NPS.

We also examined the proband two nephews:
✓ patient B.G., female, 16 years old (figure 4), present:
  • bilateral thumb nails hypoplasia
  • kyphoscoliosis
  • pectus carinatus
  • hyperextensible joints
  • elbow abnormalities and
  • left knee luxation of hypoplastic patella.

✓ Results
✓ Clinical evaluation of the proband B. M. (50 years old) revealed (figure 1):
  • Short stature (-2.9SD)
  • Microcephaly (-2.9SD)
  • Spinal and chest wall problems (large chest, dorsal kyphosis, increased lumbar lordosis,)
  • Limitation of elbows extension
  • Lateral placement of bilateral small patella
  • Bilateral dysplastic on fingers nails and toenails

✓ Radiological changes of iliac wings, elbow and knee abnormalities: small patella, narrow iliac bones, bilateral iliac horn (figure 3)

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Conclusion

✓ The diagnosis of NPS in our patients was based on clinical examination (typical major features) correlated with radiological investigation.

✓ Genetic counselling is important for this patients and the risk of recurrence is 50%.

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