

FAMILIAL CASES OF NAIL-PATELLA SYNDROME (NPS)



L.I.BUTNARIU¹, M. VOLOSCIUC², M. BURLACU², E. BRAHA¹, L. CABA¹, M. PANZARU¹,
R. POPESCU¹, M. COVIC¹, E.V. GORDUZA¹

1. "GR. T. POPA" UNIVERSITY OF MEDICINE AND PHARMACY, DEPARTMENT OF MEDICAL GENETICS, IASI, ROMANIA
2. "SAINT MARY" CHILDREN HOSPITAL, IASI, ROMANIA

Introduction

✓ **Nail Patella Syndrome (NPS)** is an hereditary osteo-oncodyplasia.

✓ (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 births²
- ❖ 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 case (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.

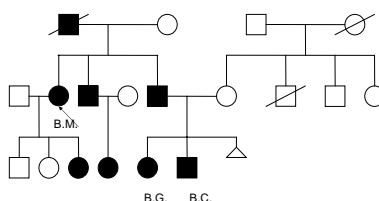


Figure 2

✓ The diagnosis was based on clinical examination correlated with radiological investigation.

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**

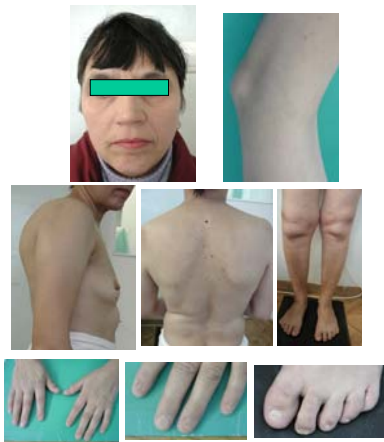


Figure 1 (B.M.)

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



Figure 3

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.**

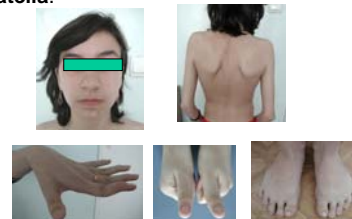


Figure 4 (B.G.)

▪ **patient B.C.**, male, 13 years old (the brother of B.G.) who presents:

- ✓ **luxation of right patella and**
- ✓ **bilateral thumbnails hypoplasia.**

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 recurrency is 50%.

Contact:

Genetic Center, Saint Mary Children Hospital, Iasi, Romania
Email: lacrybutnariu@yahoo.com
Ph/ fax: + 040232272754