Nail-patella syndrome

Nail-patella syndrome is characterized by abnormalities of the nails, knees, elbows, and pelvis. The features of nail-patella syndrome vary in severity between affected individuals, even among members of the same family.

Nail abnormalities are seen in almost all individuals with nail-patella syndrome. The nails may be absent or underdeveloped and discolored, split, ridged, or pitted. The fingernails are more likely to be affected than the toenails, and the thumbnails are usually the most severely affected. In many people with this condition, the areas at the base of the nails (lunulae) are triangular instead of the usual crescent shape.

Individuals with nail-patella syndrome also commonly have skeletal abnormalities involving the knees, elbows, and hips. The kneecaps (patellae) are small, irregularly shaped, or absent, and dislocation of the patella is common. Some people with this condition may not be able to fully extend their arms or turn their palms up while keeping their elbows straight. The elbows may also be angled outward (cubitus valgus) or have abnormal webbing. Many individuals with nail-patella syndrome have horn-like outgrowths of the iliac bones of the pelvis (iliac horns). These abnormal projections may be felt through the skin, but they do not cause any symptoms and are usually detected on a pelvic x-ray. Iliac horns are very common in people with nail-patella syndrome and are rarely, if ever, seen in people without this condition.

Other areas of the body may also be affected in nail-patella syndrome, particularly the eyes and kidneys. Individuals with this condition are at risk of developing increased pressure within the eyes (glaucoma) at an early age. Some people develop kidney disease, which can progress to kidney failure.

Frequency

The prevalence of nail-patella syndrome is estimated to be 1 in 50,000 individuals.

Causes

Mutations in the *LMX1B* gene cause nail-patella syndrome. The *LMX1B* gene provides instructions for producing a protein that attaches (binds) to specific regions of DNA and regulates the activity of other genes. On the basis of this role, the LMX1B protein is called a transcription factor. The LMX1B protein appears to be particularly important during early embryonic development of the limbs, kidneys, and eyes. Mutations in the *LMX1B* gene lead to the production of an abnormally short, nonfunctional protein or affect the protein's ability to bind to DNA. It is unclear how mutations in the *LMX1B* gene lead to the signs and symptoms of nail-patella syndrome.

Inheritance Pattern

Nail-patella syndrome is inherited in an autosomal dominant pattern, which means one copy of the altered gene in each cell is sufficient to cause the disorder. In most cases, an affected person inherits the mutation from one affected parent. Other cases may result from new mutations in the *LMX1B* gene. These cases occur in people with no history of the disorder in their family.

Other Names for This Condition

- Fong disease
- hereditary onycho-osteodysplasia
- hereditary osteo-onychodysplasia
- Osterreicher syndrome
- pelvic horn syndrome
- Turner-Kieser syndrome

Diagnosis & Management

Genetic Testing Information

- What is genetic testing?
  /primer/testing/genetictesting
- Genetic Testing Registry: Nail-patella syndrome
  https://www.ncbi.nlm.nih.gov/gtr/conditions/C0027341/

Research Studies from ClinicalTrials.gov

- ClinicalTrials.gov
  https://clinicaltrials.gov/ct2/results?cond=%22Nail-Patella+Syndrome%22+OR+%22nail-patella+syndrome%22

Other Diagnosis and Management Resources

- GeneReview: Nail-Patella Syndrome
  https://www.ncbi.nlm.nih.gov/books/NBK1132

Additional Information & Resources

Health Information from MedlinePlus

- Health Topic: Glaucoma
  https://medlineplus.gov/glaucoma.html
- Health Topic: Kidney Diseases
  https://medlineplus.gov/kidneydiseases.html
- Health Topic: Nail Diseases
  https://medlineplus.gov/naildiseases.html

Genetic and Rare Diseases Information Center

- Nail-patella syndrome
  https://rarediseases.info.nih.gov/diseases/7160/nail-patella-syndrome

Additional NIH Resources

- National Eye Institute: Glaucoma
  https://nei.nih.gov/health/glaucoma/

Educational Resources

- MalaCards: nail-patella syndrome
  https://www.malacards.org/card/nail_patella_syndrome
- Merck Manual Professional Version
  https://www.merckmanuals.com/professional/pediatrics/connective-tissue-disorders-in-children/nail-patella-syndrome
- Orphanet: Nail-patella syndrome
  https://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=2614
- Resource list from the University of Kansas Medical Center
  http://www.kumc.edu/gec/support/nail_pat.html
- University of Michigan Kellogg Eye Center
  https://www.umkelloggeye.org/conditions-treatments/nail-patella-syndrome

Patient Support and Advocacy Resources

- American Kidney Fund
  http://www.kidneyfund.org
- National Kidney Foundation
  https://www.kidney.org/
- National Organization for Rare Disorders (NORD)
  https://rarediseases.org/rare-diseases/nail-patella-syndrome/

Clinical Information from GeneReviews

- Nail-Patella Syndrome
  https://www.ncbi.nlm.nih.gov/books/NBK1132

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28Nail-Patella+Syndrome%5BMAJR%5D%29+AND+%28nail-patella+syndrome%5BTIAB%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1080+days%22%5Bdp%5D

Catalog of Genes and Diseases from OMIM

- GLAUCOMA 1, OPEN ANGLE, A
  http://omim.org/entry/137750
- NAIL-PATELLA SYNDROME
  http://omim.org/entry/161200

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