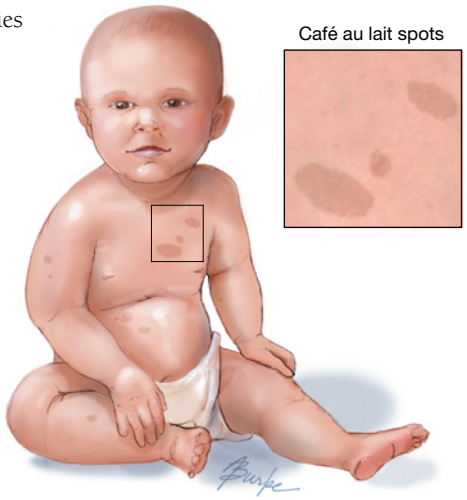
Neurofibromatosis

Neurofibromatosis (NF) is a genetic disorder causing skin abnormalities and tumors that form on nerve tissues. These tumors can be small or large and can occur anywhere in the body, including the brain, spinal cord, large nerves, or smaller nerves. NF affects persons of both sexes and all racial groups. There are 2 types of neurofibromatosis, called NF1 and NF2. These are 2 distinct disorders that are caused by **mutations** (changes) in different genes. NF1 is also referred to as von Recklinghausen disease and is a rather common genetic disease, affecting approximately 1 in 3000 individuals. Some patients who have NF1 only display characteristic skin abnormalities such as **café au lait spots**, which are flat, **hyperpigmented** (darker than surrounding skin) areas. Other patients can have severe physical complications such as **malignant** (cancerous) tumors or have mental retardation. NF2 is much more rare than NF1, affecting less than 1 in 30 000 individuals, usually becoming apparent in the late teens, and typically causing hearing loss and problems with balance due to tumors on nerves to the ears. The November 18, 2009, issue of *JAMA* includes an article about a recently described condition called Legius syndrome in which patients have skin pigment abnormalities indistinguishable from NF1. This Patient Page is based on one published in the July 16, 2008, issue of *JAMA*.



SIGNS AND SYMPTOMS OF NF1

- Café au lait spots are already visible at or shortly after birth.
- Freckling in skin folds, for instance the armpit or inguinal (groin) region
- **Neurofibromas**—soft bumps on or under the skin that are tumors arising on or along nerves
- Weakness, numbness, tingling, or other symptoms may be present if the NF tumors compress the spinal cord or large peripheral nerves.
- Visual difficulties in children can indicate a tumor of the visual pathway.
- Bone deformities, including a bowed lower leg or **scoliosis** (curved spine)
- Learning disability and mental retardation are commonly associated with NF1.

DIAGNOSIS AND GENETICS

About 50% of NF patients have an affected parent, but in 50% the mutation occurs spontaneously in a family with no previous history of NF. The diagnoses of NF1 and NF2 are based on clinical symptoms. The NF1 diagnosis can usually be made before the age of 6, whereas the symptoms of NF2 often only arise in the late teens. Since the 2 genes that cause NF1 and NF2 have been discovered, genetic testing is also available.

TREATMENT

Because of the wide range of symptoms and complications that can arise in NF1 and NF2, patients should be monitored by a team of specialists. There is currently no cure for NF1 or NF2. Surgical treatment is aimed at alleviating the symptoms that arise when NF tumors compress nearby bodily tissues and can cause damage to those tissues or organs. **Chemotherapy** (anticancer drugs) may also be offered when the tumors associated with NF are malignant. This occurs in less than 10% of persons who have NF.

FOR MORE INFORMATION

- National Institute of Neurological Disorders and Stroke
  www.ninds.nih.gov
- American Academy of Pediatrics
  www.aap.org
- Neurofibromatosis Inc
  www.nfinc.org
- The Children's Tumor Foundation
  www.ctf.org

INFORM YOURSELF

To find this and previous JAMA Patient Pages, go to the Patient Page Index on *JAMA*'s Web site at www.jama.com. Many are available in English and Spanish. A Patient Page on the basics of genetics was published in the March 19, 2008, issue.

Sources: National Institute of Neurological Disorders and Stroke, National Human Genome Research Institute, American Academy of Pediatrics, Neurofibromatosis Inc

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