

Neurofibromatosis Early Bedside Diagnosis, starting from its Inherited Real Risk.

By Sergio Stagnaro

Neurofibromatosis 1 and 2, also known as Von Recklinghausen disease, genetic conditions part of neuro-cutaneous disorders that affect the skin and the nervous system, causing tumours to grow along nerves, in the skin and brain. These tumours are usually benign.

There are two main subtypes of neurofibromatoses: Neurofibromatosis type 1 (NF1), which accounts for about 90% of all cases of neurofibromatosis, and Neurofibromatosis type 2 (NF2), which is much less common, especially in children.

Neurofibromatosis is generally an inherited disorder (autosomal dominant), but it can be caused by spontaneous mutation in up to 50% of cases. Once the change in genes happens, it can be passed on to the next generation (1-3).

According to Quantum Biophysical Semeiotic, the Inherited Real Risk of NF is always detected in all others of diseased children.

NF clinical symptomatology

Neurofibromatosis type 1 (NF1) is the most common type, affecting about one in 3,000 births. Neurofibromatosis type 2 (NF2), which is much less common, has different symptoms and is caused by changes in different genes, so is covered separately.

Neurofibromatosis type 1 is a condition characterized by changes in skin coloring, pigmentation, and the growth of tumors along nerves in the skin, brain, and other parts of the body. (1-3) According to the traditional semeiotic, the signs and symptoms of this condition vary widely among affected people.

Beginning in early childhood, almost all people with neurofibromatosis type 1 have multiple *café-au-lait* spots, which are flat patches on the skin that are darker than the surrounding area. These spots increase in size and number as the individual grows older. Freckles in the underarms and groin typically develop later in childhood.

Most adults with neurofibromatosis type 1 develop neurofibromas, which are benign tumors that are usually located on or just under the skin. These tumors may also occur in nerves near the spinal cord or along nerves elsewhere in the body.

These tumors, which usually develop in adolescence or adulthood, are called malignant peripheral nerve sheath tumors.

People with neurofibromatosis type 1 also have an increased risk of developing other cancers, including brain tumors and cancer of blood-forming tissue. (1-3). In fact, I have observed that individuals involved by NF are almost always positive for Oncological Terrain.

During childhood, benign growths, called Lisch nodules, often appear in the colored part of the [eye](#) iris. Lisch nodules do not interfere with vision. Some affected individuals also develop tumors that grow along the optic nerve. These tumors, which are called optic gliomas, may lead to reduced vision or total vision loss. In some cases, optic gliomas have no effect on vision.

Additional signs and symptoms of neurofibromatosis type 1 include high blood pressure, short stature, an unusually large head, and skeletal abnormalities such as an abnormal curvature of the spine, i.e., scoliosis.

Although most people with neurofibromatosis type 1 have normal intelligence, learning disabilities and attention deficit hyperactivity disorder (ADHD) occur frequently in affected individuals.

To summarize, NF symptomatology, it is characterized by:

Café-au-lait spots in the skin. These are flat patches of light brown or coffee-colored skin. Initially, they might be present on an infant and look like freckles, but may get larger and more numerous during a child's first few years of life, up to about 7 years old. A child with NF1 is likely to have at least six or more of these spots that are larger than freckles. The spots themselves are not painful, and people who do not have neurofibromatosis can have one or two café-au-lait spots that are benign.

Lisch nodules: These are small non-cancerous growths located on the iris (the colored part) of the eyes. Lisch nodules do not cause problems with vision, but tumors might later develop in the eye. These tumors are called optic gliomas; they may or may not affect vision.

Neurofibromas: These are non-cancerous tumors that are located mostly under the skin. Neurofibromas may also grow on nerves. In a special type of neurofibroma (called a plexiform neurofibroma), there is a 5-10% risk of developing a malignant peripheral nerve sheath tumor.

High blood pressure.

Short stature.

Macrocephaly, i.e., an unusually big head.

Bone abnormalities, such as scoliosis or tibial bowing.

Learning disabilities.

Attention deficit hyperactivity disorder (ADHD).

Seizures.

Speech problems.

Many freckles under the armpit or in the groin region – called axillary or inguinal freckling.

There may be other complications associated with NF1, including vascular (blood vessel) conditions that affect the central and peripheral nervous systems--in particular, arterial narrowing called Moyamoya disease (4, 5).

The NF1 gene provides instructions for making a protein called neurofibromin. This protein is produced in many cells, including nerve cells and specialized cells surrounding nerves (oligodendrocytes and Schwann cells). Neurofibromin acts as a tumor suppressor. Mutations in the NF1 gene lead to the production of a non-functional version of neurofibromin that cannot regulate cell growth and division.

As a result, tumors such as neurofibromas can form along nerves throughout the body. It is unclear how mutations in the NF1 gene lead to the other features of neurofibromatosis type 1, such as café-au-lait spots and learning disabilities.

NF Inherited Real Risk and its Bedside Diagnosis

Disease that cannot be healed must be eliminated with its Pre-Primary and Primary Prevention: www.sisbq.org and www.semeioticabiofisica.it

The method, I've used in my ongoing clinical research on Neurofibromatosis, 1 and 2, is the Psychokinetic Diagnostic (9, 10).

Three biological systems, CNS, nerve, and skin are involved by NF. As a consequence, in diseased individual at birth, intense (1,000 dyne cm. 2) cutaneous pinching brings about either simultaneously or after 2-3 sec. of Latency Time, according to the severity of this heritable risk, Aspecific Gastric Reflex, whose intensity parallels the seriousness of underlying disorder.

Interestingly from diagnostic point of view, the absence of Skin- Aspecific Gastric Reflex excludes the NF Inherited Real Risk. In case of melanoma Inherited Real Risk, as usually in any malignancy, the Aspecific Gastric Reflex is immediately followed by Tonic Gastric Contraction (6).

Soon thereafter, the pathological data of both Ocular and Brain- Aspecific Gastric Reflex (NN: Latency Time 8 sec. exactly; Duration > 3 sec. – 4 sec.<), corroborate the NF diagnosis. Finally,. Evoked Cerebral Potential will be clearly pathological (8).

From Clinical Microangiology view-point, the most interesting event is the Activated Microcirculation, type 2, dissociated in both the parietal, temporal, occipital area, and in the skin, whose intensity is related to the seriousness of NF Inherited Real Risk (11, 12).

All mothers of children suffering from NF, even apparently healthy, are positive at its heritable risk, that has to be eliminated with Reconstructing Mitochondrial Quantum Therapy, according to Pre-Primary Prevention (12).

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