
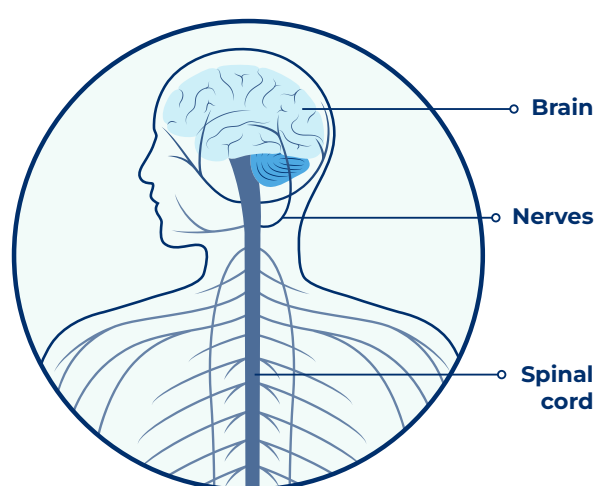


# Neurofibromatosis Type 1 (NF1) Plexiform Neurofibromas (PN)

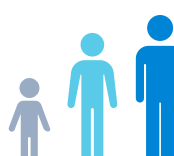
## WHAT IS NEUROFIBROMATOSIS TYPE 1?

Neurofibromatosis (NF) is a rare, progressive, genetic condition, with the most common type being **neurofibromatosis type 1 (NF1)**. NF1 can impact every organ system and be involved in the development of non-malignant (non-cancerous) tumours that may affect the brain, spinal cord and nerves.<sup>1,2</sup>

 NF1 is also one of the most common inherited disorders and is caused by a mutation or flaw in the NF1 gene. **Half** of all people with NF1 are estimated to **inherit the mutation** from a parent while the other half **have no family history**.<sup>1-3</sup>



 NF1 affects about **1 in 3,000** people worldwide.<sup>2</sup>

 NF1 is **commonly recognised in early childhood** and is a lifelong, progressive condition that affects all types of people, **regardless of gender or ethnicity**.<sup>2,4</sup>

**The severity of NF1 can vary significantly and people with the disease may not develop every symptom.<sup>5</sup>**

Initial signs of NF1 may include:<sup>5</sup>



**Flat, light brown spots on the skin** (called 'café au lait' spots)



**Soft lumps on and under the skin** (neurofibromas)

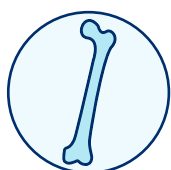
Patients with NF1 may also experience:<sup>5</sup>



**freckling in unusual places** (such as the armpits or groin area)



**optic gliomas** (tumours that develop in the cells surrounding the optic nerve)



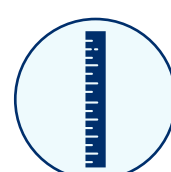
**bone issues**



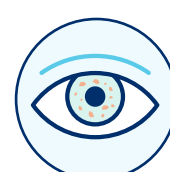
**increased head size**



**scoliosis** (back bone deformities)



**small stature**



**tiny bumps on the iris of the eye** (called 'lisch nodules')



**delayed or early puberty**



**neurocognitive differences**



**hypertension** (high blood pressure)

## WHAT IS NF1-PN?

 Up to **50%** of people

with NF1 may develop **non-malignant tumours on the nerve sheaths called plexiform neurofibromas (PN)**.<sup>5</sup>



**PN can appear anywhere inside or outside of the body.** While they are often identified in infancy or childhood, the impacts of PN can persist into adulthood. As a lifelong

condition, PN may also appear later in a person's life. Some PN can grow fast and become large, which can progressively interfere with normal physical functions.<sup>5,6,7</sup>

Although PN may start as non-malignant, **PN will later become cancerous in a small proportion (~10%) of adults** and can be associated with reduced lifespan.<sup>12,13</sup>

**PN can cause a range of physical, social, emotional and mental health problems, including:<sup>8-11</sup>**

- Pain
- Disfigurement
- Muscle weakness
- Numbness
- Breathing problems
- Bladder or bowel problems
- Visual impairment
- Anxiety (including scanxiety)
- Depression
- Social withdrawal

## HOW IS NF1-PN DIAGNOSED AND MANAGED?



**Initial diagnosis of NF1 can be complex** and may involve visits to multiple specialists that can evaluate signs, symptoms and family history as well as perform genetic testing. Upon diagnosis, **a physical examination can be conducted to understand the involvement of PN**.<sup>2,6</sup>

**An MRI (magnetic resonance imaging) of the affected body part can help determine the size and extent of the PN** on the outside and inside of the body. **Other radiology procedures may be necessary** if there is concern for the presence or development of a malignancy surrounding the nerves, including a PET (positron emission tomography) scan.<sup>2</sup>



Because NF1-PN can impact the ability to perform daily tasks, reduce quality of life and lead to additional medical complications in adulthood, **timely and continuous disease management is critical** to improving outcomes over a patient's lifetime.<sup>1,5</sup>

Content created by Alexion, AstraZeneca Rare Disease

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