Chapter 31

Neurofibromatosis

Peter Nthumba and Louis Carter

Neurofibromatosis is frequently seen in Africa. Neurofibromatosis is a genetic disorder of the nervous system with tumors on nerves. It occurs in three different types, Type 1 and 2 and Schwannomatosis. In this latter rare type there are multiple schwannomas all over the body. Simply put Type 1 is mainly peripheral and Type 2 is central though there is some overlap.

Type 1 (NF1) is also known as von Recklinghausen’s disease causes skin changes, peripheral neurofibromas, bone deformities and begins at birth and will usually be definitely evident by 10 years of age. The NF1 gene is located on chromosome 17. This type will be discussed in this chapter.

Type 2 (NF2) is associated with hearing loss, ringing in ears, poor balance, acoustic neuromas and begins in the teenage years. NF2 gene is located on gene 22. This is rarely recognized in the district hospital.

Many inherit neurofibromatosis but 30-50% of new cases arise spontaneously through mutation. These mutations may be passed on to succeeding generations.

♦ Symptoms NF1:
  - Café-au-lait spots—brown spots in the skin and 6 or more >5 mm. are diagnostic
  - Freckling in axilla or groin
  - Skin nodules—neurofibroma and 2 or more are diagnostic of NF1
  - Plexiform neurofibroma involves many nerves and maybe painful
  - Lisch nodules—neurofibromas in the iris of the eye and 2 or more are diagnostic
  - Abnormal spine development—scoliosis, kyphosis, and boy defects of skull and tibia
  - Tumor on optic nerve—optic nerve glioma
  - A parent or sibling with NF1

♦ Prognosis:
  - Often these patients have a normal life expectancy but the findings are usually progressive
  - Neurofibromas will often continue to grow
• Plexiform neurofibroma may become massive and significantly impair quality of life and may lead to psychological problems.
• Children have a higher than average risk for learning disorders.
• Malignant change is rare, <10% of patients.
• Hypertension is also seen in NF1.
• There is no cure for neurofibromatosis and nothing will limit the growth of the neurofibromas.
• Surgery rarely removes the entire tumor and after surgery, recurrence is common.

♦ Treatment:
  • Many different types of treatment have been tried but nothing has been found that is curative.
  • Radiation and chemotherapy have been tried without success.
  • Gene therapy when available will be the ultimate solution.
  • Only surgery has been found helpful but only with limited success.
  • Preoperative embolization has promise in major centers.
  • Multiple small lesions may be treated with electrocautery.

♦ Surgery:
  • Often patients want small to medium size neurofibromas, skin nodules and tags, removed because of location when on the face or neck, belt or bra line, etc. Usually these can be easily removed under local anesthesia with or without the use of electrodessication.

• Surgery may be done for large disabling and cosmetically embarrassing tumors.
• Most large lesions cannot be completely removed but only debulked.
• **Neurofibromas are very vascular and hemorrhage** may be a significant problem.

• This surgery requires excellent anesthesia and availability of blood.

• The large neurofibromas (plexiform neurofibromas) **cannot be “shelled” out** as they infiltrate the overlying skin, cross tissue boundaries and invade surrounding tissue.

• The skin often must be removed with the underlying subcutaneous tissue and the resulting defect closed with by mobilization of surrounding tissue, use of flaps or even skin grafts.

• Preop expansion of surrounding normal skin is possible before excision of the neurofibromatosis as in the case below.

![](image)

- **Scalp, expanded with a tissue expander over several weeks, was used to cover the large defect remaining after excision of the neurofibroma.**

• When neurofibromatosis involves the face, the only way to remove the mass is with near total removal of the facial skin and possible muscle and parotid excision. **This will require flap coverage.** If microvascular surgery is possible distant tissue may be used for reconstruction. Still the resulting defect may be as disfiguring as the original mass. Obviously this may lead to significant psychological issues.

• Often the neurofibroma will come out of the orbit through the supraorbital nerve. If this is the case, then the mass can be debulked with a fair result the case above. When performing surgery around the face, lidocaine with adrenaline, should be used to reduce hemorrhage. One must be careful to dilute the lidocaine to ½% so as not to overdose (Maximum amount is 7 mg/kg of lidocaine with adrenaline.) The only way to potentially remove all the neurofibromatosis in this case was to perform a craniotomy and remove the roof of the orbit—not practical where the surgery
was performed. Even then it may not be possible to remove it all. Secondary procedures are often necessary to remove residual or recurrent tumors. The patient below had 3 procedures.

Facial neurofibromas can be difficult to manage.
- With orbital involvement there is often proptosis, pulsatile eye and corneal damage secondary to exposure.
- Enucleation is the most reasonable procedure.
- Debulking may be done when one eyelid is involved but if both are involved and there is corneal damage, enucleation is the best procedure as was done for the cases below.

If only the eyelid is involved and the cornea is not damaged, debulking may be done with a good result. Repeat surgery may be necessary. See figures 5 and 6 above and figures 9-11 below.
A relatively new technique uses mesh that is used for hernia repairs. This has been shown to control the growth of disfiguring neurofibromas seen in the cheek as the case below. This “netting” procedure first described by Park from South Korea uses mesh to control recurrence of the residual mass and prevent re-drooping of the face after the initial radical but likely only palliative surgery.

The mesh provides a tight net to prevent the bulging and drooping which occurs with the force of gravity. In cases where the patient has enough normal skin that can be saved and used after wide excision of the neurofibroma, the mesh can preserve the underlying muscles of facial expression and nerves.

- The mesh is anchored to the dermal layer of the corner of the mouth, the upper lip, eyebrow, zygomatic arch and laterally to the deep temporal fascia. This gives a tight “net” around the residual tissue.
• In some patients with recurrent disease, the neurofibroma often involves the skin. These patients may need radical removal of the neurofibroma as well as much of the overlying skin. These will need flap reconstruction with deltopectoral, supraclavicular flaps (see section on perforator flaps), expanded tissue (flaps) or free microvascular flaps.
• Mesh can also be used under the flaps in these cases. An example of the netting procedure is below, the same as the eye case above.

![Fig 13](image13.png)  ![Fig 14](image14.png)  ![Fig 15](image15.png)

After neurofibroma was excised mesh was inserted and anchored to deep fascia and bone around the mass (Courtesy Dr. Peter Nthumba)

• With massive neurofibromatosis excision will not be easy because of the extent of the mass and significant hemorrhage. Just a debulking procedure on the patient’s arm below had to be terminated due to massive bleeding.

![Fig 16](image16.png)
Plexiform neurofibromatosis

• When the neurofibromatosis is limited to one nerve or one side of the extremity, debulking may be possible. The neurofibromatosis in the young girl below involved branches of the ulnar nerve. It
was resected with a long and tedious dissection under tourniquet control in order to save the nerve.

Fig 17  Fig 18
Neurofibroma in forearm and hand, along ulnar nerve, was removed first and bleeding controlled. Then tourniquet removed and remainder in arm removed. This is the same patient as seen in Figure 1 where the neurofibroma of earlobe was “completely” removed.

- When the neurofibromatosis in one extremity is massive, as in the cases below, amputation may be best.

Fig 19  Fig 20  Fig 21
Amputation was recommended in these cases

♦ Complications
- Inadequate removal but with prolonged improved quality of life
- Hemorrhage
- **Postoperative edema** is often severe and resolves very slowly and initially family members often wonder if surgery has been actually performed. This edema can occlude an airway.

**Summary**
Neurofibromatosis can be a very disfiguring and debilitating disease in Africa. The stigma must be unbearable to the unfortunate ones afflicted with this disease. It is always a very vascular. By the time these patients consult a surgeon, the disease is often far beyond easy removal. Often a few neurofibromas on the face and neck can be removed easily without significant bleeding. The use of mesh to prevent late drooping is a significant new treatment for the grotesque facial masses. Complete removal of large facial masses may require both a neurosurgeon and an ophthalmologist. Even then it will be difficult to completely remove the mass.