Definition of Neurofibromatosis

- Neurofibromatosis is a genetic disorder that disturbs cell growth in the nervous system, causing tumors to form on nerve tissue(neurofibromas).

- These tumors may develop anywhere in your nervous system

- What kind of tumors are they?

The tumors are usually noncancerous (benign) *due to suffix-oma*, but in some cases these tumors become cancerous (malignant) tumors

Epidemiology and genetics

- Autosomal dominant
- 50% of cases spontaneous mutations
- Gene located on long arm of chromosome 17 (NF1)
- Prevalence 1:4000
- First peak - 5-10 years
- Second peak - 36-50 years (75% of clinical problems due to malignancy)

Neurofibromatosis

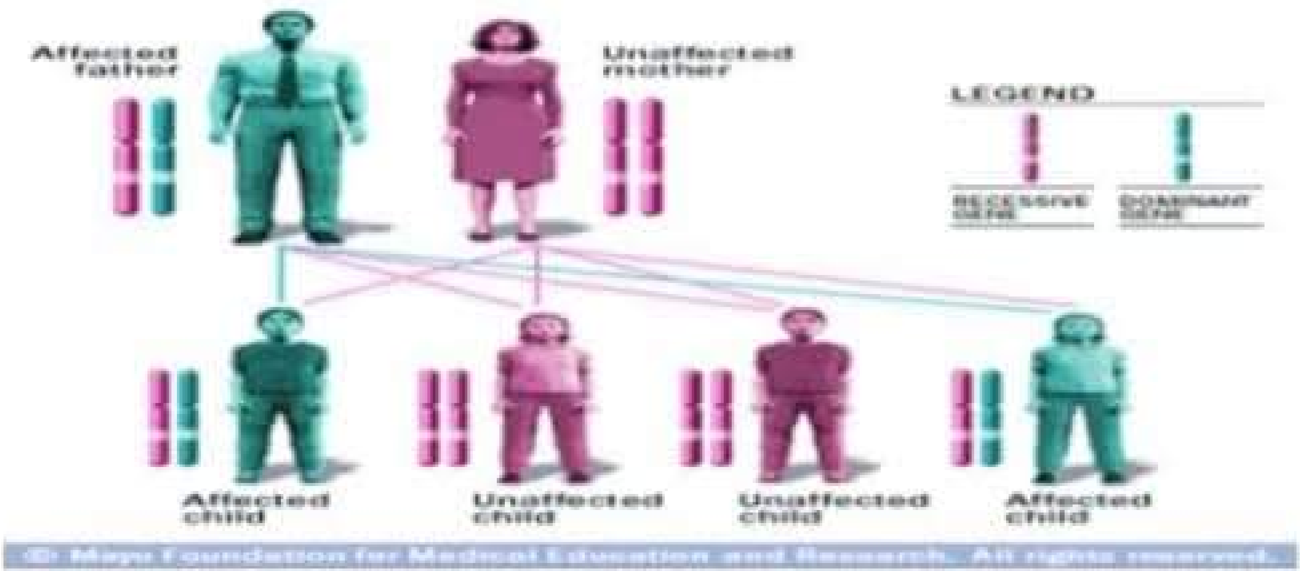
- Introduction – "Elephant Man Disease"
  - Autosomal dominant
  - Affects bone, skin, and nervous system
  - Eight clinical phenotypes
  - Two genetic disorders
- Incidence
  - Neurofibromatosis-1 (NF1), peripheral NF
    - 1 in 25-33K births
  - Neurofibromatosis-2 (NF2), central NF
    - 1 in 50-120K births
  - Segmental NF – single body region
    - Due to segmental conditional hyperexpression, mosaicism, or heterozygosity loss

Neurofibromatosis

- ETIOLOGY
  - NF1 is caused by DNA mutations located on the long arm of chromosome 17 responsible for encoding the protein neurofibromin.
  - NF2 is caused by DNA mutations located in the middle of the long arm of chromosome 22 responsible for encoding the protein merlin.

Risk factors

- The biggest risk factor is the family history, about the half is inherited
- It is an autosomal dominant
- The remaining cases result from a spontaneous genetic mutation occurs with unknown causes



NEUROFIBROMATOSIS TYPE 1

- Also known as peripheral NF, von Recklinghausen's disease.
- Neuroectodermal tumors with autosomal dominant inheritance.
- 1 person per 3500–4000 persons in the general population.
- Men and women equally affected.
- No racial predilection.
- The gene for NF-1 has been localized to chromosome 17q11.

Neurofibromatosis-I

* The most prevalent type

* multisystem genetic disorder

* Cutaneous findings, most notably café-au-lait spots and axillary freckling

* Skeletal dysplasias

* Growth of both benign and malignant nervous system tumors, most notably benign neurofibromas

Neurofibromatosis-II

* 1/25,000
* Diagnosed when 1 /4 features is present:

(1) Bilateral vestibular schwannomas

(2) Unilateral vestibular schwannoma and any 2 of the following: meningioma, schwannoma, glioma, neurofibroma, or posterior subcapsular lenticular opacities

(3) A parent, sibling, or child with NF-2 and either unilateral vestibular schwannoma or any 2 of the following: meningioma, schwannoma, glioma, neurofibroma, or posterior subcapsular lenticular opacities

(4) Multiple meningiomas (2 or more) and unilateral vestibular schwannoma or any 2 of the following: schwannoma, glioma, neurofibroma, or cataract

Neurofibromatosis

- NF1
- 1:4000 patients
- Often inherited but 30-50% occur as mutations
- 5 or more café au lait spots (some may be present at birth)
- 2 or more neurofibromas
- Most lead healthy normal lives, occasionally surgery may be required e.g. painful disfiguring lesions

- NF 2
- 1:40,000 patients
- Bilateral 8th nerve tumours
- Presents in early teens with hearing loss and symptoms of pressure on adjacent cranial nerves and structures e.g. headache, facial numbness, poor balance, tinnitus

Clinical Features

- Short statured
- Café-au-lait (CAL) spots
- Freckling
- Lisch Nodules
- Neurofibromas
- Optic gliomas

NEUROFIBROMATOSIS

Diagnosis of NF1

DIAGNOSTIC CRITERIA
Café-au-lait macules (≥ 6)
Skin fold freckling
Lisch nodules (> 2)
Optic pathway glioma
Skin neurofibromas (≥ 2) or plexiform neurofibroma
Distinctive bone abnormality
NF1 in a parent, child, or sibling

Diagnosis of NF-2:presence of **ONE** of the following criteria

Diagnostic criteria for NF2 (these include the NIH criteria with additional criteria)

Bilateral vestibular schwannomas
A first-degree relative with NF2 **AND**
- Unilateral vestibular schwannoma **OR**
- Any two of: meningioma, schwannoma, glioma, neurofibroma, posterior subcapsular lenticular opacities*
Unilateral vestibular schwannoma **AND**
- Any two of: meningioma, schwannoma, glioma, neurofibroma, posterior subcapsular lenticular opacities*
Multiple meningiomas **AND**
- Unilateral vestibular schwannoma **OR**
- Any two of: schwannoma, glioma, neurofibroma, cataract

* "Any two of" = two individual tumors or cataract.

Management

* There is no cure for neurofibromatosis.

* Patients should be routinely monitored for complications.

*Annual examinations (G&D, ophthalmo, neuro, hearing, BP, scoliosis)

* Chemotherapy, radiation therapy, or both may be used to treat cancerous tumors

* Surgery can be used to remove tumors that cause pain or a loss of function.

* Genetic counseling

NURSING MANAGEMENT:

- **Nursing assessment:**
- Obtain the detail history of patient.
- Monitor the patient for symptoms.
- Monitor the vital sign.
- Physical Assessment.
- Check the frequently intravenous site if any reaction during continuing antibiotic injection.
- Maintain intake and output chart.
- Avoid taken fat and cold drink to this type patient.
- If the patient having history of the diabetes mellitus then monitoring random blood sugar accurately and then administer insulin.
- To give details health education regarding the neurofibroma.

Nursing diagnosis

1. Acute pain and discomfort related to treatment and prolonged immobility as evidenced by oral complaint of patients and discomfort.

2. Altered nutrition level: less than body requirement related to inadequate intake of food by the patient as evidenced by weakness, fatigue.

3. Knowledge deficit related to the disease condition and recovery as evidenced by the anxiety and frequent questioning.

4. Sleep pattern disturbance related to the hospitalization as evidenced by drowsiness and discomfort.

5. Risk for injury related to seizures, disorientation and brain damage.

6. Ineffective therapeutic regimen related to lack of knowledge regarding Neurofibromatosis and its management as evidenced by frequent question prognosis.