"Health is not simply the absence of sickness." - Hannah Green

May is Neurofibromatosis Awareness Month

Neurofibromatosis

Neurofibromatosis is a genetic disorder that disturbs cell growth in your nervous system, causing tumors to form on nerve tissue. These tumors may develop anywhere in your nervous system, including in your brain, spinal cord and nerves. Neurofibromatosis is usually diagnosed in childhood or early adulthood.

The tumors are usually noncancerous (benign), but in some they may become cancerous or malignant.

Causes:

Each form of neurofibromatosis is caused by mutations in different genes.

- **Neurofibromatosis 1 (NF1):-** The NF1 gene is located on chromosome 17. A mutation of the NF1 gene causes loss of neurofibromin, which allows cells to grow uncontrolled.
- **Neurofibromatosis 2 (NF2):-** A similar problem occurs with NF2. The NF2 gene is located on chromosome 22. A mutation of the NF2 gene causes loss of merlin, which also leads to uncontrolled cell growth.
- **Schwannomatosis: -** Schwannomatosis may be associated with a mutation of the SMARCB1 gene located on chromosome 22.

Symptoms:

Neurofibromatosis 1 (NF1) usually appears in childhood. Signs and symptoms include:

- Flat, light brown spots on the skin.
- Freckling in the armpits or groin area.
- Soft bumps on or under the skin (neurofibromas).
- Tiny bumps on the iris of the eye (Lisch nodules).
- Bone deformities.
- Learning disabilities.
- Larger than average head size.
- Short stature.

Signs and symptoms of Neurofibromatosis 2 (NF2) may include:

- Gradual hearing loss.
- Ringing in the ears.
- Poor balance.

- Facial drop.
- Numbness and weakness in the arms or legs.
- Pain.
- Balance difficulties.

When to Contact a Medical Professional:

Call your health care provider if:

- You notice coffee-with-milk colored spots on your child's skin or any other symptoms of this condition.
- You have a family history of neurofibromatosis and are planning to have children, or you would like to have your child examined.

Coping and support:

Caring for a child with a chronic illness such as neurofibromatosis can be a challenge, even if the illness is mild. Greater severity of symptoms can make it even more difficult. There are many medical appointments to keep, treatments to track and instructions to follow. All of this, in addition to general concern about your child's health and anxiety over outcomes, can be overwhelming. It may help to keep in mind that many children with neurofibromatosis grow up to live healthy lives with few, if any, complications.

Prevention:

- Genetic counseling is recommended for anyone with a family history of neurofibromatosis.
- Annual eye examinations are strongly recommended.

Reference Links:

- http://www.nlm.nih.gov/medlineplus/ency/article/000847.htm
- http://www.mayoclinic.org/diseases-conditions/neurofibromatosis/basics/coping-support/con-20027728
- en.wikipedia.org/wiki/Neurofibromatosis

For any enquiry or assistance please contact: wellness@medicaretpa.co.in