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Commentary

Neurocutaneous Developmental Disorders in Children and its Diagnosis

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DESCRIPTION

Disorders classified as neurocutaneous syndromes can affect the bones, organs, skin, and spinal cord. Tumors in these tissues may develop as a result of the disorders, which are chronic diseases. In particular, they may cause to difficulties like hearing loss, seizures, and developmental problems. Specific disorders have different symptoms. Skin growths are caused on by the most common disorders in children.

The three neurocutaneous disorders which occur most frequently are:

1. Tuberous Sclerosis (TS)
2. Neurofibromatosis (NF), which includes NF1 and NF2 as well as Schwannomatosis
3. Sturge-weber syndrome

These disorders are all present at birth (congenital). They are caused by gene mutations.

Tuberous Sclerosis (TS)

The brain and retina of the eye both experience growths identified as tubers as a result. Various other body organs are also affected by tuberous sclerosis. It may have an impact on the bones, lungs, heart, kidneys, skin, and spinal cord. Also, it can cause learning difficulties, developmental delays, seizures, and intellectual disability.

Neurofibromatosis type 1 (NF1)

This is the common type of neurofibromatosis. It is also known as Von Recklinghausen's disorder. The classic symptom of NF1 is light brown color patches of pigment on the skin. These are known as cafe au-lait spots. A child may also have skin tumors that are not cancer (harmless). These are called neurofibromas. Neurofibromas are found developing on the nerves and in organs. The higher rate of brain tumors present in people with NF.

Neurofibromatosis type 2 (NF2)

Symptoms usually continue to appear between the ages of 18 and 22. Schwannomas are tumours that develop on a vestibular nerve branch. These are called as bilateral vestibular schwannomas (BVS). These 8th cranial nerve tumors can cause hearing loss, migraines, problems with facial movements, balance issues, and difficulty walking. Hearing loss in children is possible. Seizures, tumors of the membranes surrounding the brain and spinal cord (meningiomas), skin nodules (neurofibromas), and cafe-au-lait spots may also be signs of NF2.

Schwannomatosis

Schwannomas spread throughout the body as a result of this type of neurofibromatosis, but no other NF1 or NF2 symptoms are present. When a schwannoma grows large or compresses on a nerve or surrounding tissue, the predominant symptom is intense pain. Numbness, tingling, or weakness in the fingers and toes are examples of further symptoms.

Sturge-weber disease

The port wine stain, which appears on a child's face, is the disease's identifying symptom. A flat spot of skin that ranges in color from red to dark purple is called a port wine stain. It has been there since birth. It most frequently appears around or near to the eye and forehead. The birthmark leads to the growth of an excessive number of small blood vessels under the skin. On the same side of the brain as the face lesion, there may also be related brain abnormalities. Also, a child may suffer intellectual disability, physical weakness, eyesight problems, and seizures. Glaucoma, or increased eye pressure, may already present in an infant. Other body organs are unaffected by Sturge-Weber disease.

Diagnosis

- **Genetic tests:** These blood tests check for diseases that frequently run in families.
- **MRI:** This test creates images of the inside of the body using large magnets, radio waves, and a computer.
- **CT scan:** Images of the inside of the body are produced by this test using a computer and a series of X-rays. A CT scan provides more information than a regular X-ray.

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- **Electroencephalogram (EEG):** Through sticky pads (electrodes) attached to the scalp, this test records the electrical activity of the brain.
- **Eye exam:** This is done to check for retinal growths and excessive eye pressure.
- **Biopsy:** It is possible to obtain a small sample of tissue from a tumor or skin lesion. A microscope is used to check this sample.