MEDIBEATS
The Medical Bulletin

In Neurology

PHAKOMATOSES (NEURO - CUTANEOUS SYNDROME)

- Group of neuro-cutaneous disorder characterized by involvement of structure that arises from the embryonic ectoderm- typically affects CNS, the eyes and the skin.
- ***Common phakomatoses are:***- Neurofibromatosis 1 (NF 1), Neurofibromatosis 2 (NF2) , Tuberous Sclerosis, Sturge-Weber syndrome, Von- Hippel Lindau disease.
- ***Less common:***- Ataxia Telengectasia, Cowden syndrome, Klipple Trenaunay.
- **NF 1 : –**
  - ✦ Common(90% of all NF cases),
  - ✦ Always diagoned by 10 years of age,
  - ✦ Peripheral neurofibromatosis,
  - ✦ Cutaneous lesion common (>95%) -

    (Café-au-lait spot (>=6) - earliest manifestation, Freckling of armpits or groin),

    CNS lesion less common (15-20%).
- **NF 2 : –**
  - ✦ Less common (10 % of all NF cases),
  - ✦ Diagnosed- 2nd to 4th decade
  - ✦ Cutaneous lesion less common ,
  - ✦ Central neurofibromatosis,
  - ✦ CNS lesion (100%) –

    B/L vestibular Schwannomas, Meningioma,

    Ependymoma, Schwannoma of spinal nerve roots.
- ***Tuberous Sclerosis (Hard Tubers) : –***
  - ✦ Non-malignant hamartomas (mutation in TS1/ TS2 gene),
  - ✦ Skin lesion- adenoma sebaceum (angiofibroma of face), Shagreen patch (irregular flesh coloured skin), Ashleaf macules (trunk).

- Early onset seizure (80 % infantile),
- Neuro-psychiatric disorder- ADHD, Autism.
- CNS tumors – cortical and subcortical tumors, sub - ependymal nodules, white matter lesion, SEGA.
- Abdomen (renal angiomyolipoma), Thoracic (Cardiac rhabdomyoma) , skeletal system may be involved.
- Tuberous sclerosis complex- seizure, adenoma sebaceum, Mental retardation.

- ***Sturge- Weber Syndrome :-***
  - ***Clinical:-*** Child with developmental delay, seizure, macrocephaly and port wine stain on the body.
  - ***Imaging features:***- subcortical tram-track like pattern of calcification, parenchymal volume loss, enlarged choroid plexus, Calvarial hyperosteosis .
- ***Von - Hippel Lindau disease (Mnemonic – HIPPLE) :-*** H- Hemangioblastoma, I- Increased risk of RCC, P- Pheochromocytoma, P-Pancreatic lesion(cyst, adenocarcinoma), L-Liver, Renal and Pancreatic cyst, E-Eye (retinal hemangioblastoma), Endolymphatic sac tumors

Dr. Samir Kanta Datta
MD (Medicine)