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Case Study on 12-Year-Oldmale Patient with Neurocutaneous Syndrome

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CASE PRESENTATION:

HISTORY OF PRESENT ILLNESS:

A 12-year-old male child came to the hospital in paediatric department. And he has complaints of one episode of seizure which is a generalized tonic clonic seizures lasts for 15 minutes, pain in abdomen, loss of concentration and history of frothing, portical confusion for 15 minutes and tongue bite was positive. And he has a complaint of hyperpigmented areas distributed over face, back and buttock of right side. Patient had a known case of seizure activity which was occurred at the age of 8 years and the second episode at 11 years. On general examination patient was conscious and coherent. Patient was using anti-convulsant and steroids. Patient vitals were normal. On physical examination hyperpigmented areas over face, back and buttock of right side. Patient was diagnosed as seizure episode one year ago using anti-convulsant includes midazolam and phenytoin. His father-in-law has same complaints with hypopigmentation and seizures.

PAST HISTORY:

He is a known case of seizure activity at the age of 8 years and last episode was at 11 years of age.

SOCIAL HISTORY:

No significant social history.

FAMILY HISTORY:

History of same complaints in his father-in-law.

PREVIOUS ALLERGIES:

No significant allergies noted.

SURGICAL HISTORY:

No history of any past surgeries.

FAMILY HISTORY:

No significant family history.

PHYSICAL EXAMINATION:

VITALS: Temperature- afebrile, Blood pressure-100/90mmHg, Heart rate-80/min, Respiratory system – bilateral air entry positive, GRBS- 96mg/dl, SPO2- 98%room air.

GENERAL EXAMINATION:

On general examination patient has hypopigmented areas over face, back and buttock of right side.

PHYSICAL EXAMINATION:

On examination patient is conscious, coherent.

RESPIRATORY FUNCTION:

He has normal respiratory rate that is 32/min.

CARDIOVASCULAR:

He has heart rate which is regular with rhythm and there were no murmurs, wheezing sounds.

GASTROINTESTINAL:

He has per abdominal region soft and no extra growths and abnormalities were found.

Bowel and bladder were normal and appetite was normal.

LABORATORY STUDIES:

COMPLETE BLOOD PICTURE:

Haemoglobin- 9gm%

Red blood cells- 3.5mill/cumm

White blood cells- 9400cells

CT SCAN:

Low attenuation plexiform, neurofibromas along lumbosacral plexus, smaller neurofibromas are seen around urinary bladder, skin and sub-cutaneous lesions are evident, pelvic neurofibromas and targeted lesions were evident.

DIFFERENTIAL DIAGNOSIS:

- Schwannomatosis
- Sturge-weber syndrome
- Tuberous sclerosis complex
- Lichen striatus
- Linear leukoderma
- Nevus depigmentus
- Von Hippel-Lindau disease
- Neurofibromatosis (NF): Type I, Type II, and Type III
- Linear nevus syndrome
- PHACE syndrome
- Von hippelLindau syndrome
- Incontinent pigmenti

- Schimmel penning syndrome
- Phacomatosis
- CHILD syndrome

CONFIRMATORY LABORATORY EVALUATION:

DIAGNOSIS:

Based on the signs and symptoms and laboratory findings a diagnosis of **NEUROCUTANEOUS SYNDROME WITH LEFT COMPLEX SEIZURES WITH HYPOPIGMENTATION OF ITO (INCONTINENTIA PIGMENTI ACHROMIANS)** was made.

MANAGEMENT:

Neurocutaneous syndrome which is a group of congenital disorders which affects the skin, eye and the nervous system which occurs in an early childhood or adulthood. The possible symptoms include hearing loss, seizures, pain, numbness and hypopigmentation of skin with a patches. It is a lifelong condition that may cause tumours and some other developmental problems. The main treatment for neurocutaneous syndrome is based on symptomatic. Mostly it is a lifelong condition and caused tumours on the affected areas and developmental problems. And treatment will be based on the child's age, symptoms and depending on the how severe the condition. Mostly physical and occupational rehabilitation may help the child function to cure better as well as possible. At first these neurocutaneous syndrome was basically not completely known after the child birth, it will become more clearer when the child grown and develops. Laser therapy is best for cutaneous malformations and developmental problems and if seizure activity will occur best to give anti epileptics to control the seizure activity. In some cases, corticosteroids may give based on the condition of the patient. In some cases, surgery may be done to remove the tumours. Phototherapy is used in case of hypomelonois.

DISCUSSION:

ETIOLOGY AND PATHOPHYSIOLOGY:

Neurocutaneous syndrome was a rare condition which developed among one in 3000 population in which this syndrome was based on the types includes neurofibromatosis I and II. The etiology for this condition was idiopathic and depends based on the symptoms and age of the patient. The pathophysiology includes mutation of tumour suppressor gene- loss of function- uninhibited cell growth- neurofibroma development. The main clinical features for this syndrome were hearing loss, seizures, headaches. Facial pain, numbness and hypopigmentation of the skin. And the main cause for the syndrome was abnormal development of cells in embryo and characterized by the tumours in the various parts of the body and some differences in the body.The common risk factors for this can be glaucoma, visual impairment, seizures, headache, brain blood vessel defects, mental retardation, high blood pressure, delayed puberty, scohosis, bone deformities. These were the causes related to the individualised organ systems in the body. The main cause in neurocutaneous syndrome can be the genetic abnormalities. Incontinentiapigmenti can be the X linked dominant genodermatosis which is mainly characterised by the abnormalities of the tissues and the organs mainly derived from the ectoderm and neuroectoderm. It mainly involves skin, hair, teeth and nails and has neurologic, ophthalmologicanomalies. This disease is mainly caused by the mutation of IKBKG gene which is the rare disorder present in first few weeks of the age. There are four sequential stages in this condition in this they are vesicular, verrucous, hyperpigmented, hypopigmented.

DIAGNOSIS:

Diagnosis of incontinentiapigmentiis based on clinical findings. Paediatricians and dermatologists are usually the first to diagnose the disease during infant stage. Experts have established the major and minor criteria to help make the diagnosis. For the criteria for the diagnosis of incontinentiapigmenti suggests that the typical cutaneous manifestations be considered as major criteria. The diagnostic criteria for this can be six or more café-

au-lait macules, auxiliary or inguinal freckling, two or more Irish lisch nodules, two or more neurofibroma or one plexiform neurofibroma, a distinct osseous lesion such as sphenoid dysplasia, optic gliomas, bilateral vestibular schwannomas can also be the case. These were the diagnostic procedures or the diagnostic criteria related to this neurocutaneous syndrome condition.

TREATMENT:

Incontinentiapigmenti is an X-linked dominant inherited genetic disorder which is one of the groups of neurocutaneous disorders.Such types of disorders could affect the CNS, skin, eyes, teeth and skeletal system. There is no precise and particular treatment for incontinentiapigmenti. The most important aim is to halt any kind of secondary bacterial infections of skin lesions and to close observation of any other skin related problems. This should include frequent dental check-up and close monitoring by an ophthalmologist for the first few years of life.These skin lesions would usually fade away when the patient reaches adolescence or adulthood without any kind of treatment.Neurological symptoms like seizures, muscle stiffness and weakness or muscle spasms are usually treated by a neurologist with few medications like anti epileptics, anti spasmodics, anti-anxiety medications or any medical devices. Visual disturbances are treated by using corrective lenses, medications or eye surgeries in very severe cases.Dental complications can be treated by a dentist.In few cases, surgery may be done to remove tumours or extra growths that could be malignant.Complex neurofibromas can grow which can become symptomatic issuewhen they start compressing the spinal cord or airways and usually referred for surgical management. As there is high risk for optic gliomas, patients must go for regular visual evaluation, at least until they reach 6-7 years of age since optic gliomasare unlikely to develop after this age.If the patient tends to develop symptoms of an optic glioma, chemotherapy is usually recommended.Early recognition of any neurocognitive disturbances will give the patient the best opportunities to develop and function well into adulthood. It is best to do a MRI brain imaging and kidney imaging every 1-3 years in case of any CNS and renal manifestations.

PROGNOSIS:

The prognosis is usually fluctuating and it mainly depends on the extent of the involvement of the multiple organ systems apart from skin, particularly in the presence of neuro and developmental complications. Morbidity and mortality majorly are a result of opthamologic and neurologic complications, also including epilepsy visual disturbances, mental retardation. Few patients with structural brain abnormalities and seizures in newborns or neonates who are at greater risk for motor and intellectual impairment.

ENHANCED HEALTH CARE OUTCOMES:

Basically, this study tells about the professionals who worked in the proper diagnosis making like how the professionals and health care workers work as a team. This teamwork leads to proper decision making for clear and proper diagnosis. The responsibility has to be taken by every health care staff for achieving the clear and proper diagnosis. The staff who are generally involved in this process are physicians, nurses, duty medical officers, clinical pharmacists and lab technicians. These all have prominent role.in making of proper diagnosis the alterations have to be made and these all to be discussed by the staff. If there is not communication between the team then it leads to improper diagnosis then leads to improper treatment. This can lead to chronic situations. Communication can be a prominent role in making proper and clear diagnosis.

REFERENCES:

[1.] Troullioud Lucas AG, Mendez MD. Neurocutaneous Syndromes. [Updated 2023 Aug 7]. In: StatPearls [Internet]. Treasure Island (FL): StatPearls Publishing; 2023 Jan-. Available from: https://www.ncbi.nlm.nih.gov/books/NBK537001/

[2.] E.S. Roach, Neurocutaneous Syndromes,Volume 39, Issue 4,1992,Pages 591-620,ISSN 0031-3955,https://doi.org/10.1016/S0031-3955(16)38367-5.

[3.] Nitasha Klar, Bernard Cohen, Doris D.M. Lin,Chapter 27 - Neurocutaneous syndromes,Editor(s): Joseph C. Masdeu, R. Gilberto GonzálezElsevier,Volume 135,2016,Pages 565-589,ISSN 0072-9752,ISBN 9780444534859, https://doi.org/10.1016/B978-0-444-53485-9.00027-1.

[4.] Karen A. Chernoff, Julie V. Schaffer,Cutaneous and ocular manifestations of neurocutaneous syndromes,Clinics in Dermatology,Volume 34, Issue 2,2016,Pages 183-204,ISSN 0738-081X,https://doi.org/10.1016/j.clindermatol.2015.11.003.

[5.] E. Steve Roach, Diagnosis and Management of Neurocutaneous Syndromes, Semin Neurol 1988; 8(1): 83-96, DOI: 10.1055/s-2008-1041360

[6.] Charles M. Zaroff, Keren Isaacs,Neurocutaneous syndromes: Behavioralfeatures,Epilepsy&Behavior,Volume 7, Issue 2,2005,Pages 133-142,ISSN 1525-5050, https://doi.org/10.1016/j.yebeh.2005.05.012.

[7.] Harvey B. Sarnat, MD, FRCPC, and Laura Flores-Sarnat, MDView all authors and affiliations, Volume 20, Issue 8, Embryology of the Neural Crest: Its Inductive Role in the Neurocutaneous Syndromes, https://doi.org/10.1177/08830738050200080101

[8.] Marije E.C. Meuwissen, Grazia M.S. Mancini,Neurological findings in incontinentiapigmenti; a review,European Journal of Medical Genetics,Volume 55, Issue 5,2012,Pages 323-331,ISSN 1769-7212,https://doi.org/10.1016/j.ejmg.2012.04.007.

[9.] Alexander L. Berlin, Amy S. Paller, Lawrence S. Chan,Incontinentiapigmenti: A review and update on the molecular basis of pathophysiology,Journal of the American Academy of Dermatology,Volume 47, Issue 2,2002,Pages 169-190,ISSN 0190-9622, https://doi.org/10.1067/mjd.2002.125949.

[10.] Christian C. Swinney, BA, Dennis P. Han, MD, and Peter A. Karth, MD, MBA, IncontinentiaPigmenti: A Comprehensive Review and Update, https://doi.org/10.3928/23258160-20150610-09