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PATTERN AND CLINICAL PRESENTATIONS OF NEUROCUTANEOUS SYNDROMES AMONG CHILDREN ATTENDING OUTPATIENT DEPARTMENT OF ALLIED HOSPITAL

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Abstract:

Neurocutaneous syndromes are a group of diseases in which characteristic skin changes are observed in connection with central nervous system abnormalities, as a result of which there is a risk of developing epilepsy and other concomitant diseases.

***Aim:** The objective of this study is to study clinical presentations and diagnoses among children with neurocutaneous syndrome.*

***Place and Duration:** In the pediatric neurology department of Allied Hospital Faisalabad for one year duration from March 2019 to March 2020.*

***Patients and methods:** This is a cross-sectional hospital study. Fifty children with neurocutaneous syndrome were enrolled in the study from the age of 6 months to 15 years attending the hospital. Data was collected based on a designed data collection sheet, and a detailed skin examination was performed and evaluated by dermatologist.*

***Results:** The average age of the entire study group was 4.45 ± 1.96. The majority of the surveyed group belongs to the age group of 1 to 5 years, which represents 36% of the total working population. Thirty-seven patients (74%) were male and 13 patients (26%) were female. About 24% of patients showed skin complaints from all patients, with ash leaves being the most common skin lesion.*

***Conclusion:** Neurocutaneous syndromes had more than one clinical picture. Most patients with neurocutaneous disorders had the most common seizures in these children; generalized seizure was the most common in the study group. Tuberous sclerosis was the most common type of neurocutaneous disorder in our patient clinic. Dermatological examination is important for the early detection of neuromuscular disorders.*

***Key words:** neurocutaneous syndromes, skin lesions, tuberous sclerosis, dependent nodules.*

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INTRODUCTION:

Neurocutaneous syndromes are a group of diseases in which the characteristic skin changes are observed in association with central nervous system abnormalities and are ultimately exposed to the development of epilepsy. Although the features of some disorders included brain abnormalities as part of diagnostic criteria, advances in imaging and genetics have further outlined some of the rarer forms and outlined relationships with brain malformations[1-2]. This has serious implications for the procedure, since it is obvious that many, if drug resistant epilepsy is a feature, may be suitable for surgery. The most common skin lesions are: spotty patches, also called ash leaf patches, which are usually elliptical in shape, most commonly found in tuberous sclerosis, hemangiomas (sometimes called fibrous adenomas; formerly known as sebaceous adenoma), which usually include face painting regions, Shagreen patches most often visible above the lower trunk, characteristic brown fibrous plaque on the forehead, which may be the first and most easily recognizable TSC feature appreciated during physical examination of affected newborns and infants, spots in the au lait cafe which are discolored skin lesions occurring in neurofibromatosis and tuberous sclerosis and spirals and streaks also present in Ito hypomelanosis[3-4]. A characteristic feature of the TSC heart (tuberous sclerosis complex) is rhabdomyoma, a benign tumor that often occurs in the form of many lesions; uterine neuromas are one of the most common pediatric heart cancers. Most babies and children with cardiac rhabdomyemia also have TSCs[5-6]. However, rhabdomyomas are not a common finding in children with TSC found in 31% of children with tuberous sclerosis. Neurocutaneous syndromes associated with significant mortality and morbidity include uncontrolled epilepsy, a cerebrovascular accident, learning and behavioral difficulties, and brain malformations with greater impact on family and physician. No specific treatment for neuromuscular syndrome has been defined[7-8]. Management includes genetic counseling and early identification of treatable conditions or complications, electrophysiological test and brain imaging without value in asymptomatic patients. Many studies recommend such studies in all symptomatic patients, such as vision loss or disorders, proptosis, symptoms and signs of increased intracranial pressure

PATIENTS AND METHODS:

This Descriptive, observational, cross-sectional and hospital work was conducted at a pediatric neurology department Allied Hospital Faisalabad for one year duration from March 2019 to March 2020**.** The study involved 50 patients between the ages of 6 months and 15 years who were diagnosed with neurocutaneous syndromes

Admission Criteria

All children between the ages of 6 months and 15 years who meet the criteria for the diagnosis of specific neurocutaneous syndromes are eligible for this study. The study included people whose trainers agreed to take part in the study.

Exclusion criteria

• Over 15 years.
• Parents / guardians refused to participate in this study.

Data collection tools

Pre-verification form prepared for this study, personal data, sociodemographic data, details of complaints have been registered, wooden lamp examination, electroencephalography (EEG) features, magnetic resonance imaging (MRI).
All patients studied received extensive medical history, including a detailed history of current symptoms such as convulsions and developmental history. The history of autistic symptoms, hyperactivity symptoms, and similar family history such as epilepsy, mental retardation or global developmental delay are explained. A full clinical examination (general, systematic and detailed neurological examination) and dermatological examination were performed. Radiologists examined MRI and / or brain computed tomography. EEG was applied to patients who had epileptic seizures.

Ethical issues

Informed consent was obtained from all parents or guardians included in this study. Written consent was obtained from the hospital administration. Ethical approval was obtained.

Data Management and Statistical Analysis

Collected data was analyzed using the Social Sciences Statistical Package (SPSS) version 16. Data on numerical values were expressed as (mean ± SD). A p value <0.05 was considered statistically significant.

RESULTS:

Descriptive data of patients studied

Age and gender

The average age of the study group was 4.45 ± 1.96. The age is from 6 months to 15 years. The majority of the study group belonged to the age group of 1 to 5 years, which represents 36% of the entire study population. The minimum age range is 10-15 years, 8 patients make up 16% of the total. Thirty-seven patients (74%) were male and 13 patients (26%) were female. The ratio of men to women 3: 1 (Table 1 and Fig. 1).

Table 1 Age distribution among study population complaining among the working group

Age group in years	No. of patients	Percentage
6 month - 1 year	13	26
1-5 years	18	36
6-10 years	11	22
11-15 years	8	16
Total	50	100

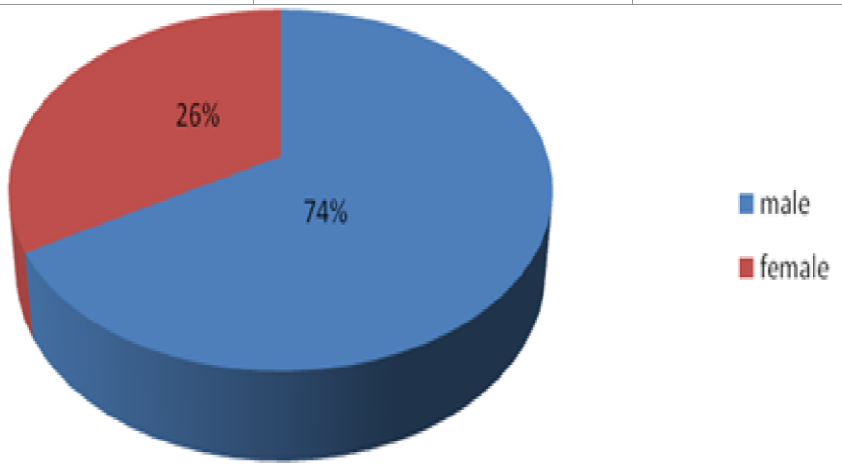

Figure 1 Sex distribution among study group

The most common complaint was seizure in 26 children (52%). Skin changes were noted in 12 cases (24%), abnormal conjunctiva in 5 children (10%), and delayed development and abnormal head size complained of 8% and 4%, respectively. A patient with tuberous sclerosis showed a headache at the age of 12 (2%) (Fig. 2).

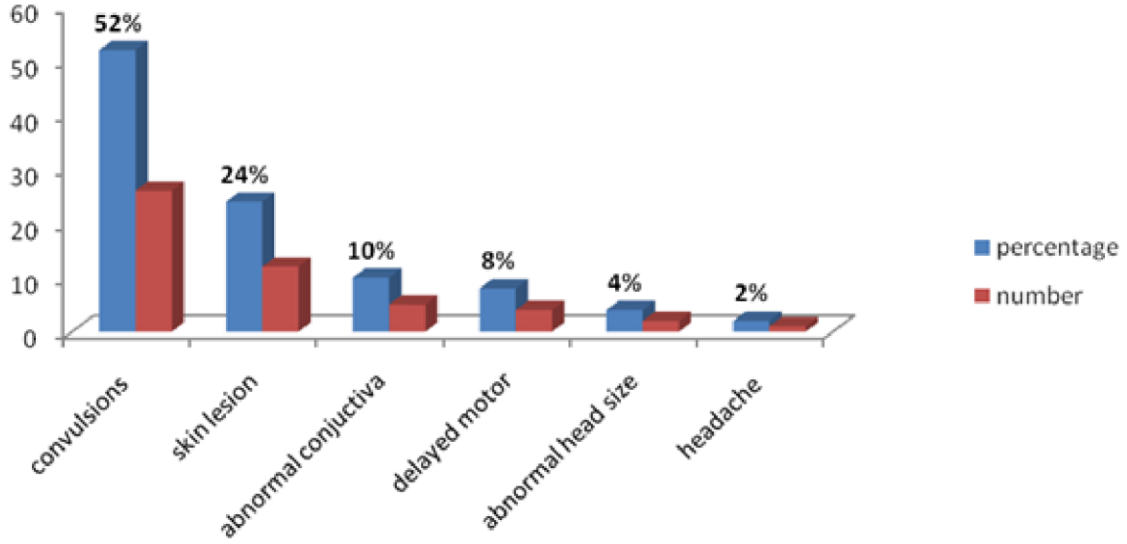

Figure 2 Frequency of the presenting complains among study population (n=50)

Convulsions were the presenting complains in 21 (80%) with children tuberous sclerosis. No convulsions have been reported in cases of telangiectasia and neurofibromatosis ataxia. About 44% of children with seizures experienced the first episode from six months to one year of age. General seizures are the most common type reported in two-thirds of patients (65.4%). Focal convulsions (23.1%) were detected and secondary generalized in 3 patients (11.5%). Among the general seizures, myoclonic seizures, i.e. infantile spasm, were the most common type and were reported in 17 (65%) patients. They were all children with tuberous sclerosis. All patients with Sturge-Weber syndrome were diagnosed with focal seizures, four patients with focal seizures, tuberous sclerosis (Table 2, Figures 3 and 4).

Table 2 Percentage of Convulsions among Specific Neurocutaneous

Syndromes	Convulsions among study group		
	Yes	No	Total
Neurofibromatosis	0	10	10
Tuberous Sclerosis	21	1	22
Sturge -Weber Syndrome	3	0	3
Hypomelanosis of Ito	2	4	6
Ataxia telangiectasia	0	9	9
Total	26	24	50

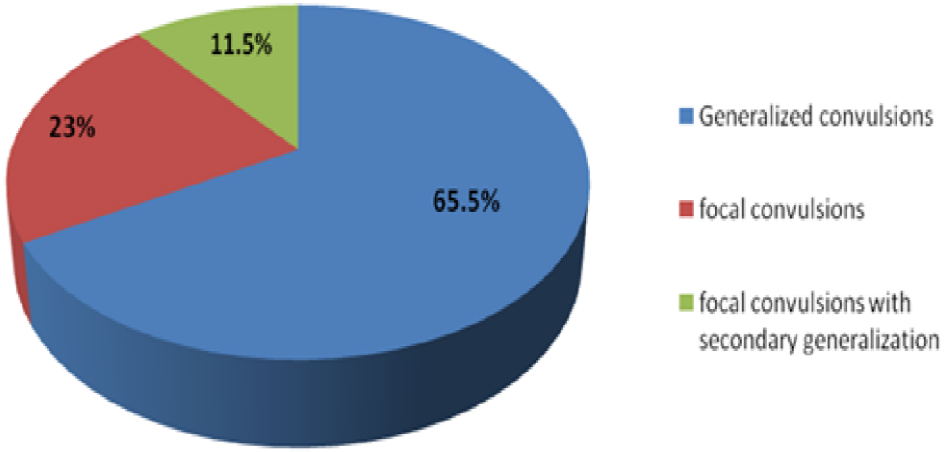

Figure 3 Percentage of the convulsions among study group

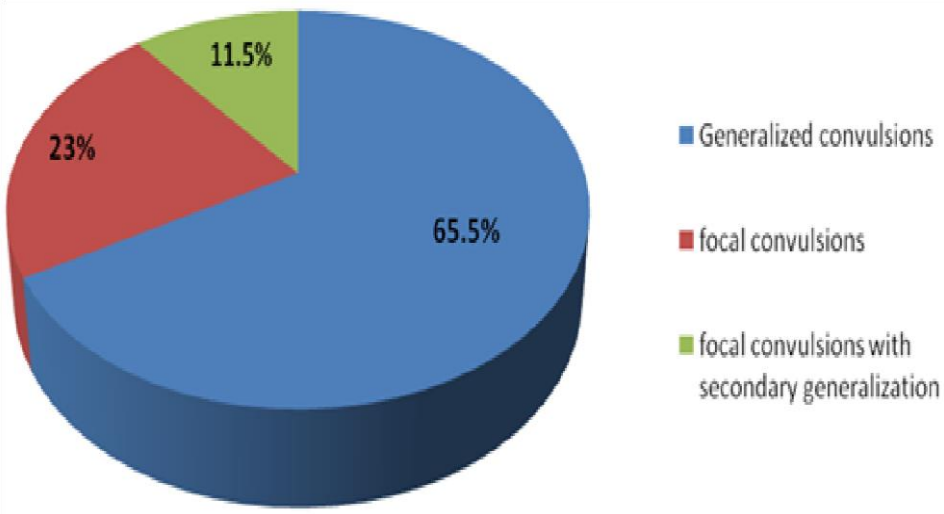

Figure 4 Distribution of the types of convulsion among study group

Skin symptoms in the study group

About 24% of patients who complain of skin lesions from all patients, white ash leaf spots were found in 15 patients. Seven children took place in the au lait cafe, five patients had neurofibromatosis and two patients had tuberculosis. Shaded patches in 9 patients, facial angiomatosis in 7 patients, spiral and wrinkles in 6 patients, ophthalmic wine stain in 3 patients and plexus neuroma in one patient with type 1 neurofibromatosis. Tuberous was a white leaf spot in 15 patients, 30%, Shagreen patch in 9 patients and facial angiotosis in 7 patients (Table 3).

Table 3 Skin lesions findings of fifty patients of the studied group (n=50)

Skin lesions	Number	Percentages
Hypo pigmented patches, ash leaf-shaped white macules	15	30
shagreen patches	9	18
Adenoma sebaceous	7	14
café au alit patches	7	14
Whorls and streaks	6	12
Birth mark-port wine stain	3	6
Neurofibroma	3	6
Total	50	100

Syndrome diagnosis

In the study group, 22 patients (44%) were diagnosed with tuberous sclerosis. Another common patient is type 1 neurofibromatosis, diagnosed in 10 patients representing the study population (20%). Ataxia telangiectasia and Ito hypomelanosis were found to be 18% and 12%, respectively. The Sturge-Weber syndrome rate was the lowest, at 6%. Regarding the distribution of different diagnoses by sex, there was a significant male dominance in all teams, but this correlation was not statistically significant (Table 4 and Figure 5).

Table 4 shows sex distribution among different study population (n=50)

Syndromes	Sex		
	Male	Female	Total
Neurofibromatosis	7	3	10
Tuberous sclerosis	16	6	22
Sturge Weber syndrome	2	1	3
Hypomelanosis of Ito	5	1	6
Ataxia telangiectasia	7	2	9
Total	37	13	50
P=0.08 insignificant			

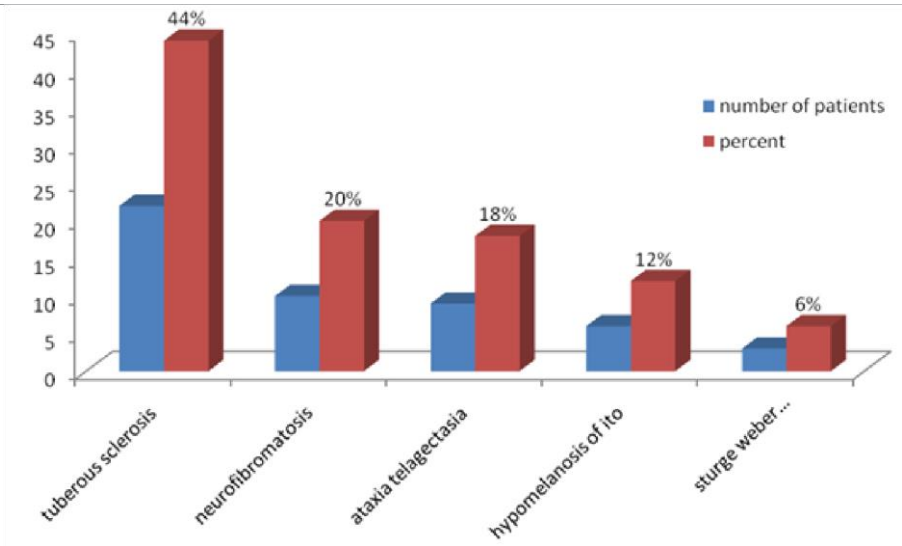

Figure 5 Distribution of the neurocutaneous syndrome among study group

DISCUSSION:

Neurocutaneous syndromes include a heterogeneous group of diseases characterized by abnormalities in both the skin and the central nervous system. Some of them can be diagnosed after birth, while others do not cause symptoms later in life. Although neurocutaneous syndromes cannot be cured, treatment can help control symptoms and emerging health problems. The average age is 4.45 ± 1.96 years and is almost similar to other studies[9-10]. The average age was less than the results of the Smirniotopoulos study, with an average age of 10 ±

1.45 years, because their work covered a wide range of age groups. In this study, the most common complaint was seizures in 52% of children studied. This result was consistent with the results obtained by other researchers who found seizures in 56% of their studies. In Kalinina's study, skin symptoms, e.g. white ash leaf spots and au lait cafe spots, were the most common presentation - 45%. These differences between tests from initial presentations may be related to the intensification of clinical presentations, because we recommend patients with skin symptoms to seek medical advice later than neurological symptoms. In this study, we could not find seizures in neurofibromatosis and it was similar to the Dahan study. The age of onset of the first seizures ranged from 6 months to 12 years and was similar to another study[11-12].

According to our study, in most cases of neuromuscular syndrome, seizures were general (65%) and the results were similar to other seizure studies with a variable rate between different studies, this change may depend on the number of populations studied in the study. The result is not consistent with other studies where the most common types of seizures are complex partial types. The difference in seizure types can be attributed to the severity of brain involvement or other independent factors affecting the younger group of people of working age. In tuberous sclerosis, all children complained of 66.7% generalized seizures and focal in 33.4% of focal sclerosis, seizures began in the first years of life and this is consistent with other studies[13-14]. In our results, we observed that in most patients with Ito melanosis, generalized seizures developed according to the results achieved by Smirniotopoulos. Almost all three cases of Sturge-Weber syndrome in our study had seizures that were difficult to control.

The authors noted that there was no seizure in patients with type 1 neurofibromatosis in the study group; a similar discovery was obtained by another person[15].

CONCLUSION:

Neurocutaneous syndromes have had many clinical presentations, most patients with neuromuscular disorders have epileptic seizures, most commonly occurring in these children; generalized seizures were the most common in the study group. Tuberous sclerosis was the most common type of neurocutaneous disorder in our patient clinic. Dermatological examination is important for the early detection of neuromuscular disorders. Skin discoveries, skin discolorations (spots of white ash leaves) were the most common changes and assisted in early diagnosis.

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