ORIGINAL ARTICLE

Skin Manifestations of Neurocutaneous Syndromes, Among Sudanese Children Attending Outpatient Clinic of Soba University Hospital

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Abstract :

Background: The neuroccutaneous syndromes are a group of diseases where characteristic skin lesions are seen in association with abnormalities of the central nervous system, and by consequence are at risk of developing epilepsy and other co morbidities. *Aim and Objectives:* The objective of this study was to list skin manifestations among Sudanese children with neuroccutaneous syndrome, attending outpatient clinic of Soba University Hospital, 2015. *Material and Methods:* This is a cross sectional hospital based study, fifty children with neuroccutaneous syndrome, were included in the study, and aged between 6 months to 15 years, from April 2014-April 2015, the data was collected by designed check sheet, and detailed skin examination was done and reviewed by dermatologist. *Results:* The mean age of the whole study group was 4.45 ±1.96 years. The majority of study group were in the age group 1-5 years constituting 36% of total population of the study. Thirty seven patients (74%) were males and 13 patients were females 26%, male, 24 % of patients presented complained of skin lesions from total patients, the most common skin lesion is ash leaves. *Conclusion:* Skin manifestations, hypo pigmented skin patches (ash leaves) was the commonest lesion, and help in early diagnosis.

Keywords: Neuroccutaneous syndromes**;** Sudanese; skin lesion; Tuberous sclerosis; subependymal tubers

Introduction:

The neuroccutaneous syndromes are a group of diseases where characteristic skin lesions are seen in association with abnormalities of the central nervous system, and by consequence are at risk of developing epilepsy**.** Although, the features of some disorders have included abnormalities of the cerebrum the diagnostic criteria, advances in imaging and genetics have further delineated some of the rarer forms and outlined associations with malformations of cerebral development. This has major implications for management as it is apparent that many, if drug resistant epilepsy is a feature, may be suitable for surgery [1, 2].

The most common skin lesions are: Hypo pigmented maculae, also known as ash-leaf spots, which are usually elliptic in shape most commonly found in tuberous sclerosis, angiofibroma (sometimes called fibro adenomas; previously called adenoma sebaceous), which typically involve the malar regions of the face, Shagreen patches, seen most commonly over the lower trunk, distinctive brown fibrous plaque on the forehead, which may be the first and most readily recognized feature of TSC to be appreciated on physical examination of affected neonates and infants, café auliat patches which are depigmented skin lesions found in neurofibromatosis and tuberous sclerosis and whorls and streaks also present in hypomelanosis of Ito [3, 4].

The characteristic cardiac feature of Tuberous Sclerosis Complex (TSC) is rhabdomyomas, a benign tumor that often presents as multiple lesions; cardiac rhabdomyomas are one of the most common pediatric cardiac tumors [5].

Most infants and children who have cardiac rhabdomyomas also have TSC However; rhabdomyomas are not a universal finding in children with TSC, found in 31 % of children with tuberous sclerosis [5].

The neuroccutaneous syndromes associated with considerable mortality and morbidity, including uncontrolled epilepsy, cerebrovascular event, learning and behavioral difficulties, brain malformations with greater impact on family and clinician.

There is no specific treatment for neurocutaneous syndrome. Management includes genetics counseling and early identification of treatable conditions or complications, electrophysiological test and brain imaging of no values in asymptomatic patients. Many studies recommend such investigations in all symptomatic patients such as visual loss or disturbance, proptosis, symptoms and signs of increased intracranial pressure [6-9].

Sudan is a country with high consanguinity rate which may increase the incidence of such syndromes among the population.

The objective of this study was to demonstrate dermatological findings in neurocutaneous syndromes among Sudanese children attending outpatient clinic of Soba University Hospital, 2015.

Material and Methods:

Study design:

Descriptive cross sectional, observational hospital based study.

Study area:

This study was conducted at a pediatric neurology outpatient clinic of Soba university hospital, which twice a week and the number of patients seen per clinic day is around 50 -60 patients. Soba university hospital is reference hospital for all paediatric neurological cases from the country.

Study period:

The study was conducted in one year (April 2014 to the end of April 2015).

Study population:

This study included 50 patients whose age ranged from 6 months to 15 years attending the pediatric neurology outpatient clinic at Soba University Hospital and diagnosed as having neuroccutaneous syndrome.

Inclusion criteria:

All children aged 6 month to 15 years, who fulfilled the criteria for diagnosis of specific neurocutaneous syndromes, were eligible for this study.

Exclusion criteria:

Children with age more than 15 years

Parents / caregiver if refused to participate in this study

Data collection tools:

Designed check sheet prepared before hand for this study was used for data collection which included personal information, socio demographic data, detailed presenting complains, detailed skin examination including; wood light, Electroencephalography (EEG) features, Magnetic Resonance Imaging (MRI) findings.

Sample size

A total of 50 patients were enrolled.

Methods:

All patients were subjected to clinical history including detailed history of the presenting symptoms like seizures and developmental history. Data on autistic symptoms, hyperactivity symptoms, and presence of epilepsy, mental

retardation or global developmental delay were also collected and family history of the neurocutaneous syndromes was recorded. Detailed skin examination was done of all patients, which was reviewed by a dermatologist.

Ethical consideration:

Informed consent was obtained from all parents or caregivers included in this study; written approval was obtained from hospital administration. Ethical clearance was obtained from research committee in Sudan Medical Specialization Board (SMSB).

Data management and statistical analysis:

The collected data were analyzed using the Statistical Package for Social Science (SPSS) for windows version 16. The data for numerical values was expressed in (Mean ± Standard division SD). *P*- value ⩽ 0.05 was considered statistically significant.

Results:

Descriptive data of the studied patients

Age and sex

The mean age of the whole study group was 4.45 ±1.96 years. The age range was between 6 months and 15 years. The majority of study group were in the age group was between 1 year - 5 years constituting 36% of total population of the study. The least age range were between 10 years -15 years ,they were 8 patients constituting 16 % of the total, thirty seven patients (74%) were males and 13 patients were females (26%), male : female ration 3:1. (Table 1, 2) (Fig. 1, 2).

Table 1: Age Distribution among Study Population

Age Group	No. of Patient	Percent
6month <1 year	13	26.0
1 year -5 year	18	36.0
6 year -10 year	11	22.0
11year - 15 year	8	16.0
Total	50	100.0

Table 2: 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 value = 0.08 insignificant

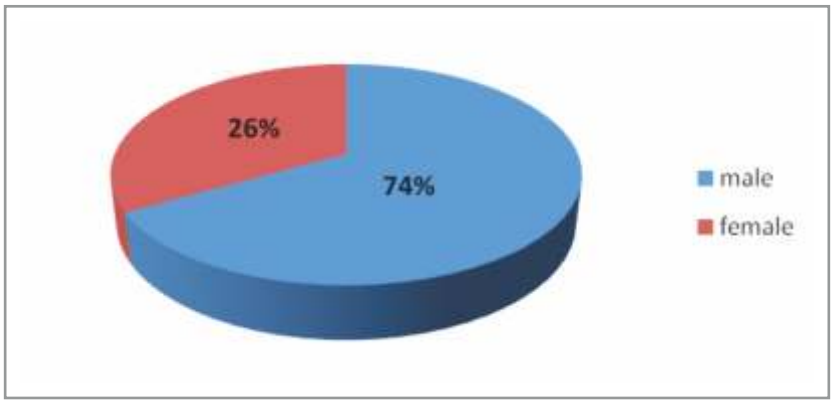

Fig. 1: Distribution by Gender Patients with Neurocutaneous Syndromes

Skin manifestations among studied group

About 24% of patients presented complained of skin lesions from total patients, ash leaves were found in 15 patients. 7 children had café auliat patches, 5 patients were children with neuro-fibromatosis and 2 patients with tuberous sclerosis. Shagreen patches found in 9 patients, facial angiomatosis in 7 patients, whorls and streak in 6 patients, port wine stain opthalamicin three patients, plexiform neuroma in one patient with neurofibromatosis type 1. The main of the skin lesions in tuberous sclerosis was ash leaves found in 15 patients constituting (30 %), Shagreen patch in 9 patients, and facial angiomatosis in 7 patients (Table 3, 4) (Fig. 3).

Table 3: Dermatological Manifestations among Patients With Neurocutaneous Syndromes

Skin lesions	Number	Percent
Hypo pigmented patches, Ash leaves	15	30
Shagreen patches	9	18
Adenoma sebaceous	7	14
Café au lait patches	7	14
Whorls and streaks	6	12
Birth mark-port wine stain	3	6
Neurofibroma	3	6
Total	**50**	**100.0**

Table 4: Dermatological Manifestations among Patients with Tuberous Sclerosis

Dermatological manifestations	Number of patients	Percent
Hypo pigmented patches, Ash leaves	15	30
Shagreen patches	9	27
Adenoma sebaceous	7	21
Café au alit patches	2	6
Total	**33**	**100.0**

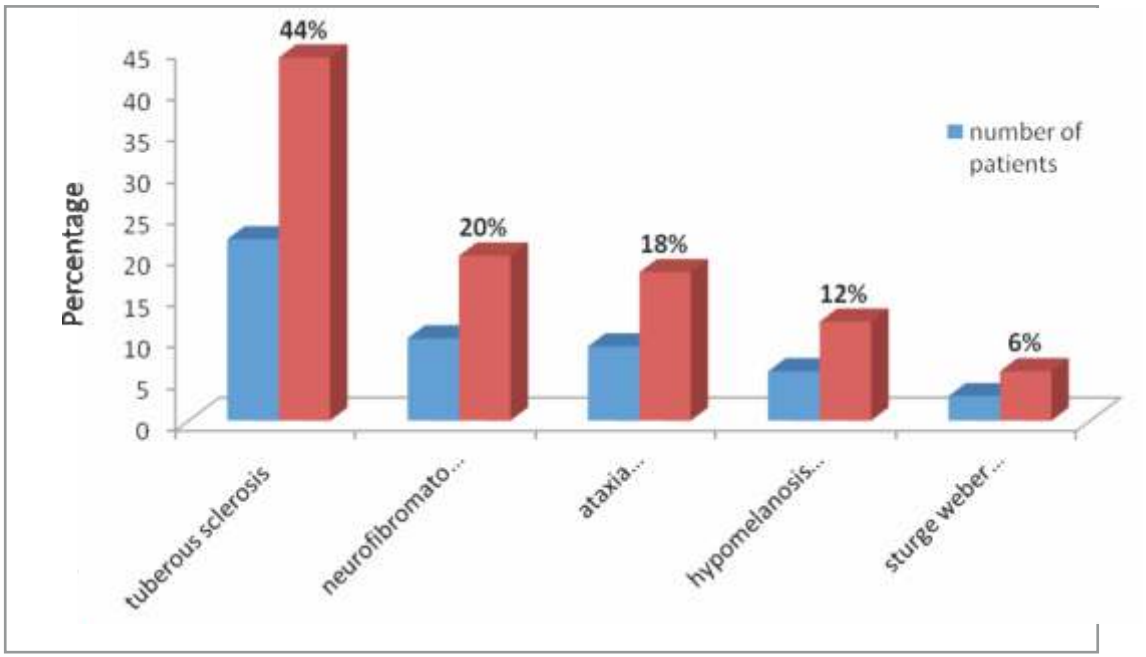

Fig. 2: Patterns of the Neurocutaneous Syndromes among Studied Patients

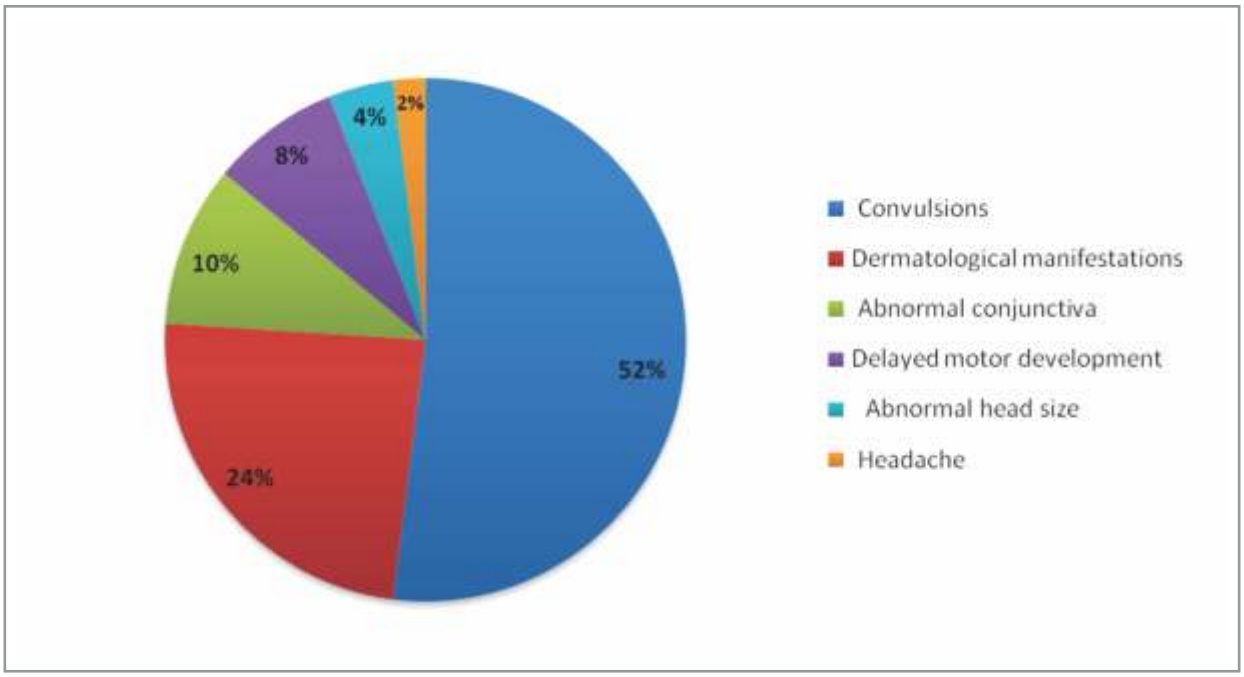

Fig. 3: Main Clinical Presentations among Studied Patients (n= 50)

Discussion:

Neurocutaneous syndromes include a heterogeneous group of disorders characterized by abnormalities of both the skin and central nervous system. While some can be diagnosed at birth; others do not produce symptoms until later in life. Although, neurocutaneous syndromes cannot be cured, treatment can help to manage symptoms and health problems that occur [3, 4, 7, 10-14].

The mean age was 4.45 ± 1.96 years which was nearly similar to other studies, Aziz, Abdulrahman, Purkait *et al*. [15-17], a lower than reported by Smirniotopoulos, Murphy, where mean age was 10 ± 1.45 years as their study included a wide range of age group [18].

In this study skin manifestations second most common clinical presentation following convulsions and in accordance with other studies Aziz, Abdelrahamin [15-17] and against two other studies Kalinina *et al.*, Salvolini *et al*., where skin manifestations e.g. hypopigmented patches was commonest presentation found in 45%, 55% respectively, this variations between studies with regard to initial presentations could be related to severity of clinical presentations as we proposed patients with skin manifestations seek medical advice later than neurological symptoms, or may be due to environmental genetic or other independent factor [22, 23].

Regard to skin manifestations, study reveals that ash leaves were commonest skin manifestations of neurocutaneous syndromes in this study. This in accordance with many other studies (Aziz, Beck *et al*., and Cross) [15, 19-21] and lower percentage in studies done by Aziz, Salvolini *et al* respectively [15, 22, 23].

In tuberous sclerosis, the most common cutaneous manifestations is ash leaves detected in this study ;presenting as initial manifestation with convulsions and this help in early diagnosis and was emphasized with other studies Abdelrahamin *et al.*, Dahan, Zaroff [16, 23-26]. Contrary to the other studies result, Shagreen patches and angiofibroma were common, Kalinina, Elster and Rose [21, 23, 27-30]. This variation was due to age group of study population. In majority of cases of neurofibromatosis were presented with café auliat patches, this was similar to the results of other studies, Beck, Hanno [18].

Conclusion:

Skin examination is important, for early detection and diagnoses of various neurocutaneous syndromes. They have done in the study from 6 month of age onwards, it will be better if they include earlier age group also.

Recommendations

Skin examination is important, for early detection and diagnosis of various neurocutaneous syndromes. Skin examination of other family members is essential part of evaluation of child with neurocutaneous syndromes.

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