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Original Article

Patterns of Neurocutaneous Syndromes in Upper Egypt

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ABSTRACT

Background: Neurocutaneous Syndromes [NCS] are rare congenital disorders manifesting at different ages with central nervous system and cutaneous abnormalities. its incidence below 1:2,000 individuals in the general population.

Aim of the Study: This study aims to determine different clinical patterns of NCS in Upper Egypt.

Patients and Methods: This is a cross section study included 80 patients with NCS selected from patients attending dermatology, pediatrics and neurology outpatient clinics of Al-Azhar University Hospital [Assiut], from 1st May 2021 till 30th June 2022. All patients enrolled in this study were subjected to complete history, dermatological and neurological examination. Computed tomography [CT], magnetic resonance Magnetic resonance imaging [MRI] and electroencephalogram [EEG] were done in some cases.

Results: Dermatological manifestation of neurofibromatosis [NF] was café au let macule [CALM], Axillary freckles and Neurofibroma. Tuberous sclerosis [TS] had Angiofibroma, Ash leaf and Shagreen patches. Ataxia telangiectasia [AT] had ocular and cutaneous telangiectasia. Xeroderma pigmentosa [XP] had Photo-sensitivity, solar lentigen and xerosis. Neurological manifestation of TS and NF was mental delay followed by Epilepsy. AT had ataxia, gradual motor regression. XP had moderate mental delay and the majority had ataxia, SWS had epilepsy, hemiplegia and mild mental.

Conclusion: NF is the most common NCS then TS, AT, Sturge-Weber, Xeroderma pigmentosum, and hypomelanosis of Ito, Linear Nevus Syndrome [LNS] and dyschromatosis symmetrica hereditra are the least. CALM is the most common dermatological manifestation in NF and TS, mental delay is the most common neurological manifestation. Port-Wine Stains [PWS] presents in all cases of SWS and epilepsy is the most common neurological manifestation. All cases of XP record Photosensitivity, solar lentigen, xerosis and mental delay, dry eye and conjunctival injection.

Keywords: Neurocutaneous; Ataxia-telangiectasia; Port wine stain; Sturge-weber syndrome; Xeroderma pigmentosa; Neurofibromatosis.

INTRODUCTION

Neurocutaneous disorders [NCS], also referred to as phacomatoses, are congenital disorders manifesting at different ages with central nervous system [CNS] and cutaneous abnormalities. These disorders are regarded as rare diseases, with incidence below 1:2,000 individuals in the general population [1]. While phenotypically and genetically diverse, they are united by their origins in defects of the developing primitive embryonic ectodermal tissue, which gives rise to both the skin and nervous system. Most NCS are classified as single-gene disorders, they may have autosomal dominant, autosomal recessive, or X-linked inheritance patterns [2].

NCS disorders include Neurofibromatosis 1 [NF1], Tuberous Sclerosis [TS], Sturge Weber Syndrome [SWS], von Hippel-Lindau disease [VHL], Pascual-Castroviejo type II syndrome [PHACE syndrome], Ataxia telangiectasia [AT], Linear Nevus Syndrome [LNS], hypo melanosis of Ito and Incontenintia Pigmenti [IP], Xeroderma pigmentosa [XP]. Among them; the most common disorders are NF, TS complex and SWS. These lifelong disorders generally present during infancy and early childhood [3].

NF type 1 is diagnosed upon the following diagnostic criteria: [Two of them must be met for the diagnosis of NF type 1]: Six or more cafe-au-lait spots, greater than 5 mm prepubertal and greater than 15 mm post-pubertal. Two or more neurofibromas or one or more plexiform neurofibroma. Axillary or groin freckling. Optic glioma. Two or more Lisch nodules. Sphenoid dysplasia, dysplasia or thinning of long bone cortex and First-degree relative with NF type [4].

TS is diagnosed upon the following diagnostic criteria: [Two major criteria or 1 major and 2 or more minor criteria must be met for the diagnosis of TS complex [TSC]] the major criteria are: Three or more hypopigmented macules of at least five mm of diameter, Three or more angiofibromas or a fibrous cephalic plaque, Two or more ungual fibromas. Shagreen patch. multiple retinal hamartomas, cortical dysplasias. Sub ependymal nodules. Sub ependymal giant cell astrocytoma, cardiac rhabdo-myosarcoma, Lymphangioleiomyosarcomatosis and Two or more angiomyolipomas. Minor criteria are: Several small hypopigmented lesions grouped together, also called "confetti" skin lesions, Three or more dental enamel pits, Two or more intraoral fibromas, A retinal achromic patch, multiple renal cysts, nonrenal hamartomas [5].

The aim of this work is to determine different clinical patterns of NCS in Upper Egypt.

PATIENTS AND METHODS

This is a cross section study included 80 patients with NCS selected from patients attending dermatology, pediatrics and neurology outpatient clinics of Al-Azhar University Hospital [Assiut], from 1st May 2021 till the end of 30th June 2022. All patients enrolled in this study were subjected to complete history, dermatological and neurological examination. CT, MRI brain and EEG were done in some cases.

The parents of all patients give a written consent form for agreeing their children to participate in the study. The study was carried out after being approved by the local ethics committee of the faculty of medicine, Al-Azhar University, Assiut, Egypt.

All patient with sign and symptoms of NCS were included; while, patients who refuse to share in the study and patient with pigmentary and neurological disorders other than NCS were excluded.

Data collection: Complete history taking was obtained from patients and their parents, complete dermatological examination of lesions was done for all patients with determining its shape, color, number, size, site with photo taking for all skin lesions by camera honor 9X Lite. Neurological assessment [complete neurological history, full neurological examination] and ophthalmological assessment. CT and MRI brain were done in some cases.

The collected data were analyzed using the Statistical Package for Social Science [SPSS] for windows version 24. The data for numerical values was expressed in [Mean ± Standard division SD], while Median and Interquartile range [IQR] for non-parametric numerical data. Frequency and percentage were done for non-numerical data.

RESULTS

Ages ranged from 6 days to 55 years, 49 [61.25%] patients were males and 31 [38.75%] were females. The majority of patients 58 [72.5%] were from rural regions, there are Positive family history in 51 [63.75%] patient and Positive consanguinity 35 [43.75%] patient. The mean age of cases with NF was 19 ± 16

years with range [4 mo. - 55y]. There are 20 males [58.8%] and 14 females [41.18%]. Patients came from rural areas are [79.4%] and those came from urban areas are [20.6%] of NF cases. Positive family history in 67.65%, and Positive consanguinity in 64.71% of NF cases.

Table [1]: Demographic Characteristic of the studied patients with NCS

General parameters		Total [n=80]
Age: [years]	Mean ± SD	14.07 ± 12.57
	Range	12 [6 d -55 y]
Sex, No. [%]	Male	49 [61.25%]
	Female	31 [38.75%]
Residence, No. [%]	Rural	58 [72.5%]
	Urban	22 [27.5%]
Positive family history		51 [63.75%]
Positive consanguinity		35 [43.75%]

Table [2]: Pattern of the NCS according to final diagnosis

Syndromes	No. [%]
Neurofibromatosis	34 [42.5%]
Tuberous Sclerosis	22 [27.5%]
Ataxia telengiectasia	11 [13.75%]
Sturge Weber syndrome	6 [7.5%]
Xeroderma pigmentosum	4 [5%]
Hypomelanosis of Ito	1 [1.25%]
Linear Nevus Syndrome	1 [1.25%]
Dyskeromatosis symmetrica hereditra	1 [1.25%]
Total	80 [100%]

Table [3]: Clinical characteristics of studied patient with NF

	Neurofibromatosis [n=34]
Dermatological manifestation	
Neurofibroma	18 [52.94%]
Axillary freckle	30 [88.24%]
Café au let macule	34 [100%]
Giant melanocytic nevus	[2.9%]
Neurological manifestation	
Mental delay	9 [26.47%]
Epilepsy	3 [8.8%]
Others	
Optic glioma	1 [2.9%]
Bilateral congenital glaucoma	1 [2.9%]
Iris lisch	19 [55.88%]

Table [4]: Clinical characteristics of studied patient with TS

	Tuberous Sclerosis [n= 22]
Dermatological manifestation	
Hypo pigmented patches or Ash leaves	20 [90.9%]
Shagreen patches	16 [72.73%]
Angiofibroma	21 [95.5%]
Neurological manifestation	
Mental delay	16 [72.7%]
Epilepsy	8 [36.36%]
Autism	6 [27.27%]
Attention-Deficit Hyperactivity Disorder	2 [9.1%]
Others	
Hamartoma related to optic disc	1 [4.55%]
Multiple renal cyst	1 [4.55%]
Subungual fibroma	2 [9.1%]

Table [5]: Clinical characteristics of studied patient with Sturge Weber syndromes [SWS]

		Sturge Weber syndrome [n=6]
Dermatological manifestation	Port-Wine Stains	6 [100%]
Neurological manifestation	Mental delay	2 [33.3%]
	Epilepsy	5 [83.3%]
	Hemiplegia	3 [50%]
Others	Glaucoma	5 [83.3%]
	Retinoblastoma	1 [16.67%]

Table [6]: Clinical characteristics of studied patient with Xeroderma pigmentosum cases

		Xeroderma pigmentosum syndrome [n=4]
Dermatological manifestation	Photosensitivity, solar lentigen and xerosis	4 [100%]
Neurological manifestation	Mental delay	4 [100%]
	Ataxia	3 [75%]
Others	Dry eye and conjunctival injection	4 [100%]

Table [7]: Clinical characteristics of studied patient with AT cases

		Ataxia telengiectasia [n=11]
Dermatological manifestation	Skin & eye telangiectasia	7 [63.6%]
	Vitiligo	1 [9%]
Neurological manifestation	Ataxia	11 [100%]
	Gradual motor regression	10 [90.9%]
	Motor delay	1 [9%]
Others	Recurrent chest infection	4 [36.3%]

Table [8]: Consanguineous marriage and similar condition among patients with Neurocutaneous Syndromes

Historical examination	Consanguineous marriage	Similar condition
Neurofibromatosis [n=34]	22 [64.71%]	23 [67.65%]
Tuberous Sclerosis [n= 22]	1 [4.55%]	16 [72.73%]
Ataxia telangiectasia [AT] [n= 11]	6 [54.54%]	8 [72.73%]
Sturge Weber syndrome [n=6]	1 [16.67%]	-
Xeroderma pigmentosum [n=4]	4 [100%]	3 [75%]
Hypomelanosis of Ito [n=1]	1 [100%]	-
Linear Nevus Syndrome [n=1]	0 [0%]	-
dyskeromatosis symmetrica hereditra [n=1]	0 [0%]	1 [100%]
Total [n=80]	35 [43.75%]	51 [63.75%]

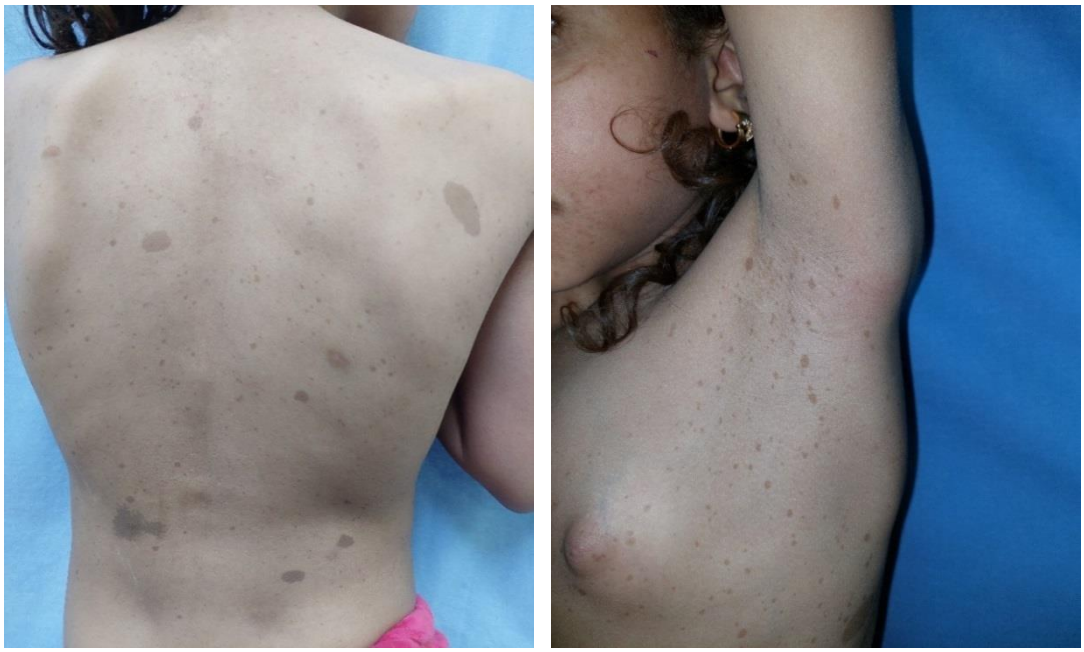

Figure [1]: Female patient 10 years old with NF1 present with CALM and axillary freckling

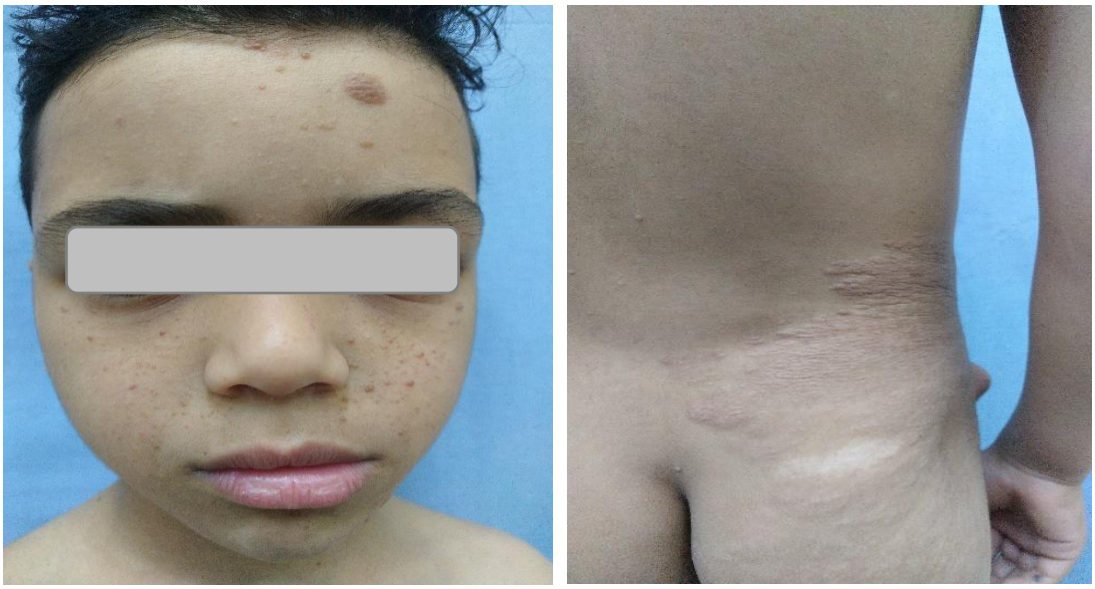

Figure [2]: male patient 9 years old diagnosed as TS present with angiofibroma at the face, shagreen patch and ash leaf macule at the back

Figure [3]: Female patient 7 years old diagnosed as AT present with ataxia and bulbar telangiectasia

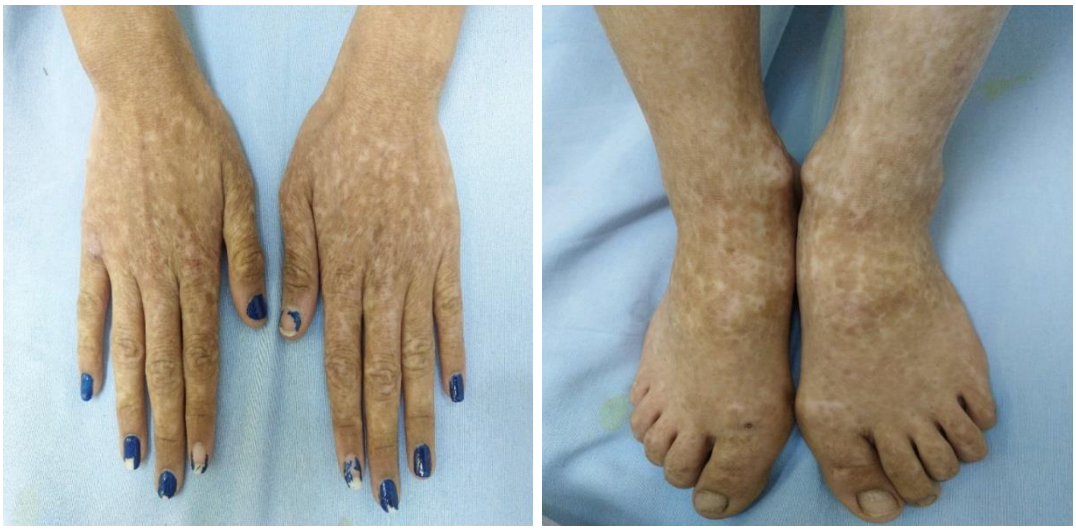

Figure [4]: Female patient 22 years old diagnosed as dyskeratosis symmetrica heredaitra present with ataxia and mottled hypo and hyperpigmentation at extremities

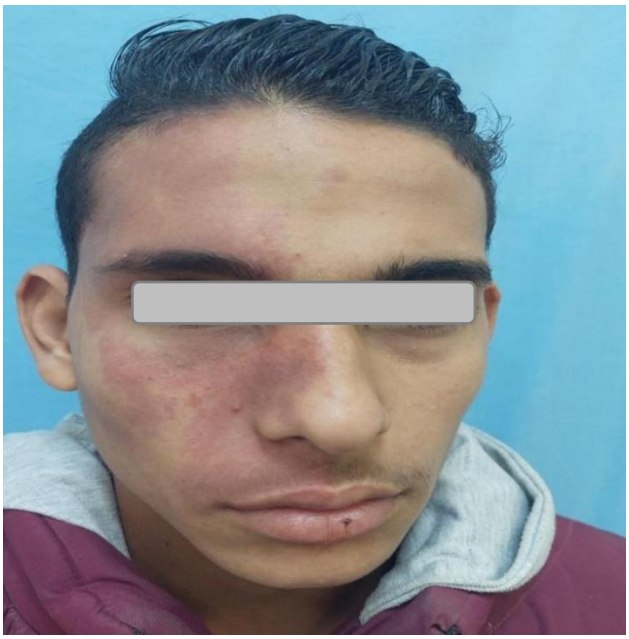

Figure [5]: Male patient 18 years old diagnosed SWS present with Port-Wine Stains [PWS] at the right side of face, glaucoma and epilepsy

DISCUSSION

NCS include a heterogeneous group of disorders characterized by abnormalities of both the skin and CNS. Treatment can help to manage symptoms and health problems that occur [7].

The current study included 80 patients diagnosed with NCS. Their ages ranged from 6 days to 55 years, with a mean ± SD of 14.07 ± 12.57 years. Out of the total, 49 patients [61.25%] were males and 31 [38.75%] were females. The majority of patients, 58 [72.5%], were from rural areas. This finding is consistent with a study conducted by **Oshi *et al.*** [8], wherein the majority of patients were males, with 37 patients [74%], while 13 patients [26%] were females, which was nearly similar to other studies such as **Sadek *et al.*** [9]. Another study mentioned that out of 27 children, 16 [59.26%] were boys, and 9 [40.74%] were girls with NCS, as observed in the study by **Kundu *et al.*** [10].

In the current study, the most common NCS was NF type 1, diagnosed in 34 patients [42.5%]. The next most common syndrome was TS, diagnosed in 22 patients [27.5%]. This was followed by AT with 11 patients [13.75%], SWS with 6 patients [7.5%], XP with 4 patients [5%], and hypomelanosis of Ito, LNS, and dyschromatosis symmetrica hereditra, each diagnosed in 1 patient [1.25%]. This contrasts with a study by **Kundu *et al.*** [10], where in 27 patients, the various forms of NCS observed were TSC, the most common found in 13 out of 27 patients [48.14%], followed by AT with 8 patients [29.62%], SWS with 4 patients [14.81%], NF1 with 1 patient [3.7%], and LNS with 1 patient [3.7%].

The current results also disagreed with another study conducted by **Sadek *et al.*** [9] in Sohag, Egypt. They found that TS was the most commonly found in 12 cases [44.45%], constituting the majority of the cases, followed by NF. XP was detected in four cases [14.82%] each. SWS cases were detected in three cases [11.11%], and lastly, IP and hypomelanosis of Ito were detected in two cases [7.40%] each. In contrast to the current study, **Goraya *et al.*** [11] conducted a survey on 87 children and indicated that TS was the most commonly diagnosed in 47 children, followed by NF type 1 and AT in 10 cases each. Six children had SWS, five had hypomelanosis of Ito, and three had Sjogren-Larsson syndrome.

According to the general characteristics among NF cases in this study, there were 20 males [58.8%] and 14 females [41.18%]. Patients from rural regions accounted for [79.4%], while those from urban regions represented [20.6%] of NF cases. A positive family history represented [67.65%], and positive consanguinity represented [64.71%] of NF cases. The current study aligns with a study conducted by **Tettamanti *et al.*** [12], which

indicated that the majority of children with NF type 1 were males [953 [53%]].

In 34 patients with NF type 1, CALM was found in all cases [100%]. Axillary freckles were present in 30 patients [88.24%], followed by neurofibromas in 18 patients [52.94%]. Among NF cases, mental delay was observed in 26.47% of cases, while epilepsy was detected in 8.8% [3 cases]. In terms of ophthalmological manifestations, the most common was Iris Lisch, found in 19 patients [55.88%]. Additionally, optic glioma and bilateral congenital glaucoma were reported in 2.9% of cases each. In a study by **Oshi *et al.*** [8], it was found that CALM appeared in 96.5% of 1063 patients, axillary and inguinal freckling in 90% of 991 patients, and neurofibromas in 78.1% of 861 patients.

According to TS, there were 14 males [63.64%] and 8 females [36.36%]. Among the TS cases, 50% of patients came from rural areas while the other 50% came from urban areas. In line with the current study, **Tomin *et al.*** [13] explained in their survey on TSC patients that although there were more boys than girls in the study sample, the difference in sample size and gender representation was not significant enough to draw conclusions regarding whether there is a gender effect on the risk of TSC.

In the current study, the dermatological manifestation of TS showed that Angiofibroma represented 21 cases [95.5%], Hypopigmented patches or Ash leaves represented 20 cases [90.9%], and Shagreen patches represented 16 cases [72.73%]. In terms of neurological symptoms, mental delay was found in 16 cases [72.73%], epilepsy in 8 cases [36.36%], while Autism and ADHD were found in 2 cases [9.1%].

A study agreed with our study, skin manifestations of TSC include facial angiofibroma, hypomelanotic macules, shagreen patches and periungual fibroma. Facial angiofibroma 12 [92.30%] was the most common finding followed by hypomelanotic macules 10 [76.90%] and shagreen patch 10 [76.90%] **Kundu *et al.*** [10] Nearly similar findings were found in different studies **Kingswood *et al.*** [14].

Regarding the current study, there was 4 males [66.67%] and 2 females [33.3%]. Patients came from rural areas were [83.3%] and those came from urban areas represented [16.67%] of SWS cases. Positive consanguinity represented 16.67% of SWS cases.

This Agrees with a study done by **Yan *et al.*** [15] on 57 patients in which the incidence of SWS in males 34 cases [59.6%] more than females 23 cases [40.4%].

Dermatological manifestation, PWS at right side was found in all cases. Neurological manifestation, Epilepsy was found in 5 cases [83.3%], Hemiplegia which was found in 3 cases [50%], Mild mental was the least and found in 2 cases 33.3%.

This agreed with a recent study done by **Rihani *et al.*** [16] on 13 patients where all studied patients had PWS [100%], the most common neurological manifestation was seizures 4 cases [31%], followed by hemiparesis 3 cases [23%].

According to our present study, there was 3 males [75%] and 1 female [25%] with XP. The mean of age of onset was 33 ± 6 years. Dermatological manifestation included photo-sensitivity, solar lentigen and xerosis. Neurological manifestations included moderate mental delay [100%], 3 cases [75%] had ataxia. Ophthalmological manifestations included dry eye and conjunctival injection [100%].

In a study done by **Rabie *et al.*** [17] in Upper Egypt, on 36 XP patients the author revealed that the incidence in females 21 [58.3%] was more than males 15 [41.7%], which disagrees with our study. All patients developed classic XP skin abnormalities including xerosis, skin atrophy, lentigines, and poikiloderma on face and extremities and Majority of patients 29 [80.5%] developed ocular symptoms, namely, photophobia, conjunctivitis, and keratitis which agreed with our study.

According to the current study, there was 7 males [63.64%] and 4 females [36.36%]. 100% came from rural areas. Positive family history represented 72.73%, and Positive consanguinity represented 54.54% of AT cases. The mean of age of onset was 57.84 ± 15 years. This agreed with a study done by **Mahadevappa *et al.*** [18] on 100 patients, the incidence in males 60 [60%] was more than females 40 [40%]. The mean age of onset of illness was 3.9 ± 2.84 years, which is lower than that presented in the current study. A positive family history was obtained in 20% this incidence is less than that

in our study and consanguinity was obtained in 60% which is near to the incidence in our study.

In our study dermatological manifestation, all cases [100%] had ocular and cutaneous telangiectasia, only one case [9%] had vitiligo. Neurological manifestation, ataxia [100%], 10 cases [90.9%] had gradual motor regression, one case [9%] had motor delay. Recurrent chest infection presented in 4 cases [36.6%].

This also agreed with **Mahadevappa *et al.*** [18] study in which also all cases had ocular and cutaneous telangiectasia and ataxia, less than 5 cases [<5%] had other dermatological manifestations such as café au lait spots, hypopigmented macules, vitiligo, and seborrheic dermatitis, recurrent sino-pulmonary infections presented in 64 patients [64%].

Conclusion: NF is the most common NCS, TS, AT, Sturge-Weber, Xeroderma pigmentosum, and hypomelanosis of Ito, LNS and dyschromatosis symmetrica hereditra are the least. CALM is the most common dermatological manifestation in NF and TS, mental delay is the most common neurological manifestation. PWS presents in all cases of SWS and epilepsy is the most common neurological manifestation. All cases of XP record photosensitivity, solar lentigen, xerosis and mental delay, dry eye and conjunctival injection.

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