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Original Article

# Pattern of Genodermatoses among Paediatric Population at a Remote Area of Jammu: Cross Sectional Prospective Study at a Tertiary Centre

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## ABSTRACT

**Background**:- Genodermatoses encompasses a group of disorders either congenital or hereditary affecting the structure and function of skin. Only few studies are available on genodermatoses so this study was conducted to characterize genodermatoses from an area remotely located with unique epidemiological and genetic population composition. Material and methods:- A cross-sectional observational prospective study was carried out at a tertiary centre of a remote area of north India. In this study which was carried out over a period of 2 years( January, 2022 -2024), all patients within the paediatric age group (0-18 years) were taken up for the study, who presented with any type of genodermatoses. Results : This was an observational based hospital study conducted over a period of 1 year. A total of 54 patients with genodermatoses were detected out of a total OPD of 18230 patients. Thus the prevalence came out to be
.29. Conclusions: we need to have more and more studies from various regions of our country on geneodermatoses which will surely facilitate the development of larges databases for early and accurate diagnoses.

***Keywords****: genodermatoses, paediatric population, keratinization disorders, bullous disorders.*

## Introduction

Genodermatoses encompasses a group of disorders either congenital or hereditary affecting the structure and function of skin. They may exclusively affect the skin or multisystem involvement may be there. The inheritance can be variable and can present either at birth or later in life. In the skin, they may have a conglomeration of symptoms affecting the colour, texture of epidermis or may involve the dermis, connective tissues or appendages. Several genodematoses can cause great mortality and ,morbidity.[1] It is imperative to have a comprehensive view of the geneodermatoses to help family cope up with the risk of recurrence in future generations and offer any symptomatic or genetic treatment if available. The relative rarity and the lack of resources for genetic testing in resource poor nations further endorses the need for such studies to establish a comprehensive databases. There has been a dearth of research studies from India as well. To the best of our knowledge, only few stdies are available on genodermatoses so his study was conducted to characterize genodermatoses from an area remotely located with unique epidemiological and genetic population composition.[2]

**Material and methods**: A cross-sectional observational prospective study was carried out at a tertiary centre of a remote area of north India. In this study which was carried out over a period of 2 years( January, 2022 -2024), all patients within the paediatric age group (0-18 years) were taken up for the study, who presented with any type of genodermatoses ,to the dermatology out patient department on their own or referred from other departments, at a tertiary care centre of peripheral area of muslim majority

population. The term genodermatoses encompassed all proven genetic and congenital disorders transferred from one or both parents to one or more of their children because of the specific abnormalities (mutations) in their genes. Moreover, in the perspective of congenital disorders we, included all dermatological conditions presenting with structural abnormalities of the skin that exist at, and usually before, birth regardless of their etiology.

A total of 52 patients with clinically proven genodermatoses were taken up for the study after proper informed consent from the patients and the parents. A detailed clinical history and examination including age of onset, family history, progression, exacerbating and relieving factors, and dermatological and extracutaneous symptoms, morphology, configuration and distribution of lesions were noted. Detailed neurological, ophthalmological, cardiological Also, epidemiological details including age, gender, marital status, ethnicity, onset of the disease, parental consanguinity, family pedigree were recorded.

Routine blood investigations including complete blood count (CBC), total leukocyte count (TLC), differential leukocyte count (DLC), erythrocyte sedimentation rate (ESR), liver function tests (LFT), renal function tests (RFT) and urine routine were done in all. Systemic evaluation including neurological, ophthalmological, cardiological were done wherever indicated. A broad categorization of the genodermatoses was also carried out to potray the basic genedermatoses etiology. All the statistical analysis was done using the using the SPSS software version 20.0 (SPSS Inc., Chicago, IL, USA). The continuous variables were measure in terms of central tendency. The categorical variables were measured in terms of percentages and absolute numbers.

**Results:**

This was an observational based hospital study conducted over a period of 1 year. A total of 54 patients with genodermatoses were detected out of a total OPD of 18230 patients. Thus the prevalence came out to be .29.

There was a male predominence with 31/54 and females 23/54. Male to female ratio was 1.2. A high degree of consanguinity was noted in 51 out of 54(98%) patients and family history of similar complaints amongst first degree relatives was seen in 12 patients.

The age of the patients ranged from new born babies to 18 years. The distribution of the patients in the various age groups is shown in table 1. In the age group of 0 – 6 years, 19 patients presented.

In the age group of 7-12 years, 20 patients reported whereas 12 patients were in the age group of 13-18 years. The broad categorization of the patients in various genodermatoses is shown in figure 1.

In our study, genetic disorders keratinization was the most common group of disorders, seen in 19/54(33.3%)patients. Icthyoses was seen in 12 patients with male preponderance, keratoderma in 2 males and females each. Among icthyoses patients, icthyoses vulgaris was commonly 5 males and 3 females. Non bullous icthyosiform erythroderma in 2 females, lamellar icthyoses in 2 males.(figure 2) In the 2 female patients, clinical and histopathological diagnoses was of aqua syringeal keratoderma. One patient, 17 years old was clinically diagnosed to be a case of Naxos syndrome.(figure 3,4)She presented with short curly hair since birth and hair growth was reduced. Also there was history of thickening of palms and soles since childhood. There was episodic shedding of nails of hands and feet. The patient further gave history of recurrent pus filled lesions turning into crusted plaques on and off. Consultation with cardiologist along with electrocardiogram, echocardiography did not show any abnormalities. Another patient, 1 year old was clinicaly diagnosed with ectodermal dysplasia as the patient had sparse scalp hair, nail dystrophy and decreased sweating. Teeth were normal.

The next common category of genodermatoses seen was vascular malformations seen in 12/54(22.2%) of the patients. Among these, haemangiomas was seen in 11 patients and klippel tranaunay syndrome in 1 patient of 3 years old.

Familial tumor syndromes and DNA instability disorders were seen in 11/54(21%) of the patients. Most common entity was neurofibromatosis seen in 6 patients, tuberous scleroses in 4 patients and xeroderma pigmentosa seen in 1 patient. Among 6 patients of neurofibromatosis, the oldest one was 11 years old and the youngest 4 years old. All patients of neurofibromatoses presented with café au-lait macules, axillary freckling and multiple neurofibromas. Patients were screened for ophthalmological and auditory examination. All patients of tuberous scleroses had angiofibromas and ash leaf macules, one patient had abnormalities on ophthalmological examination.

The next category was genetic blistering disorders seen in 7 patients. Out of these 7, six patients presented with epidermolysis bullosa simplex(EBS)(.figure 5) One patient was diagnosed with ectodermal dysplasia skin fragility syndrome(EDS/SFS). The patient underwent genetic testing where in whole exome sequencing showed homozygous single base pair deletion of plakophilin(PKP1) gene at exon 12 was detected. No other variants of unknown significance was detected. A diagnosis of EDS/SFS was confirmed. (Figure 6,7,8)

Four patients were diagnosed as suffering from genetic pigmentary disorders. Out of these, 2 were presenting as

oculocutaneous albinism and both were siblings. The other 2 were clinically diagnosed as cases of dyschromatoses universalis hereditaria.

In the miscellaneous group, one 6 month old child presented with diarrhea, periorificial rash, nail changes. Another 11 year old child presented with features of xeroderma pigmentosa. There was hypopigmentation and hyperpigmentation, atrophy, telengiectasias, actinic keratosis. One superficial basal cell carcinoma was present on the face.

Table 1 . Age and sex distribution of various genodermatoses.

| Genodermatoses | 0-6 years | | 7-12 years | | 13-18 years | | *TOTAL* | |
|---|---|---|---|---|---|---|---|---|
| | Males | Females | Males | Females | Males | Females | *Males* | *Female* |
| 1.Icthyosis(12) | 3 | 1 | 5 | 2 | | 2 | 8 | 5 |
| 2.keratoderma(4) | | | | 1 | 2 | 1 | 2 | 2 |
| 3.Ectodermal dysplasia | 1 | | | | | | 1 | |
| 4.Naxos syndrome (1) | | | | 1 | | | | 1 |
| 5.Epidermolysis bullosa simplex.(4) | 1 | 4 | | | | | 1 | 4 |
| 6.Dystrophic epidermolysis bullosa(2) | 1 | | | | | | 1 | |
| 7.Ectodermal dysplasia skin fragility syndrome.(1) | 1 | | | | | | 1 | |
| 8.Dyschromatoses universalis hereditaria | | | 1 | | 1 | | 2 | |
| 9.Neurofibromatoses | | | | 3 | 1 | 2 | 1 | 5 |
| 10.Tuberous scleroses | 1 | | 1 | 1 | 1 | | 3 | 1 |
| 11. Haemangiomas | 4 | 3 | 3 | 1 | | | 7 | 4 |
| 12.acrodermatitis enteropathica | 1 | | | | | | 1 | |
| 13.klippel trenaunay syndrome | 1 | | | | | | 1 | |
| 14.Xeroderma pigmentosa | | | | | 1 | | 1 | |
| 15.Oculocutaneous pigmentation | | | | 1 | 1 | | 1 | 1 |

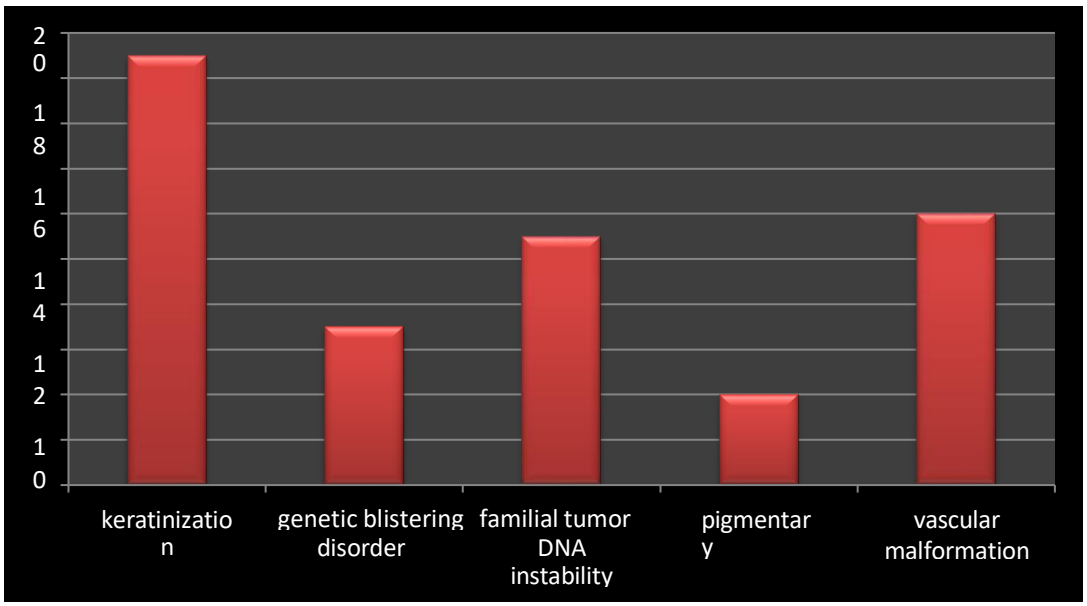


Figure 1 showing broad categorization of genodermatoses disorders

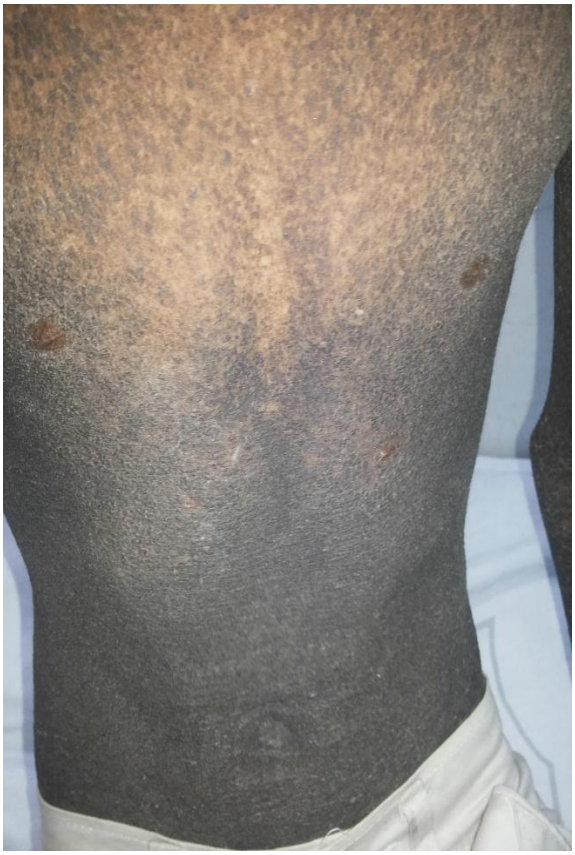

Figure 2

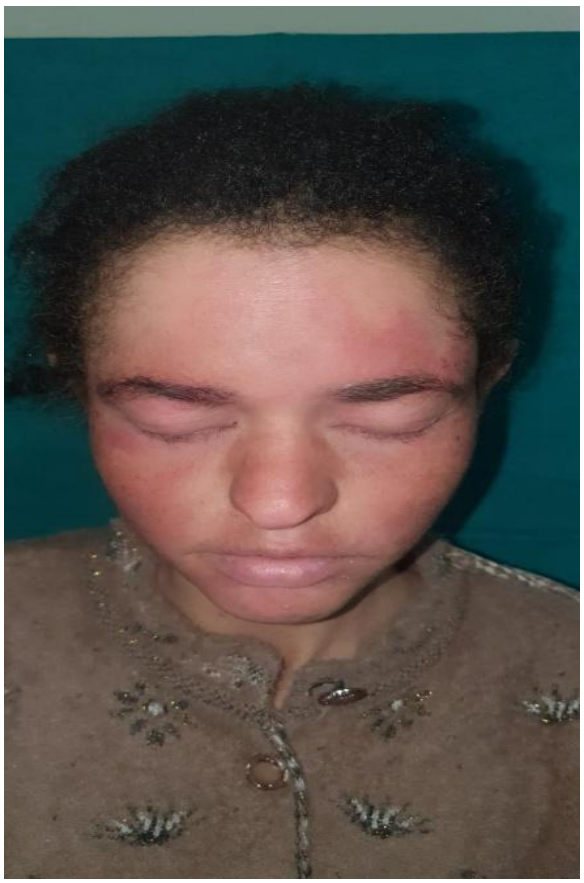

Figure 3

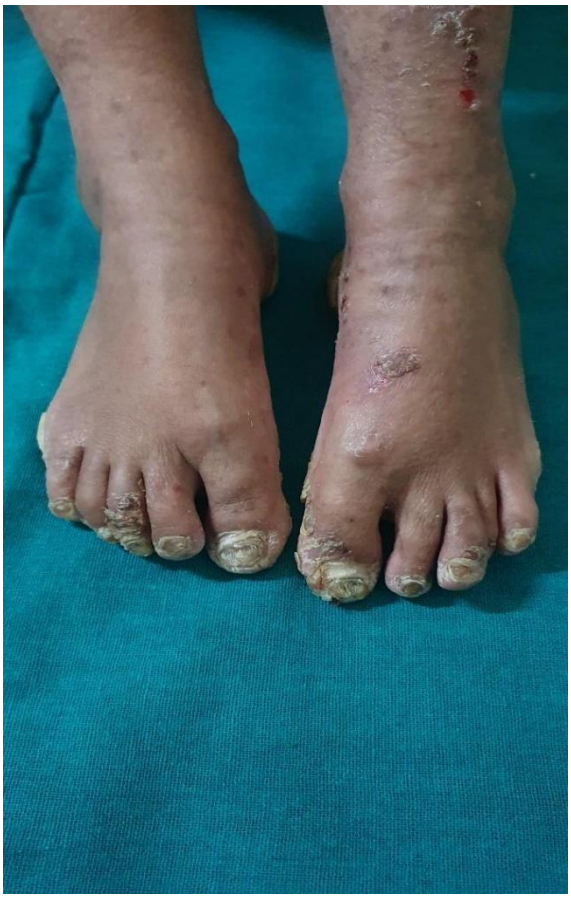

Figure 4

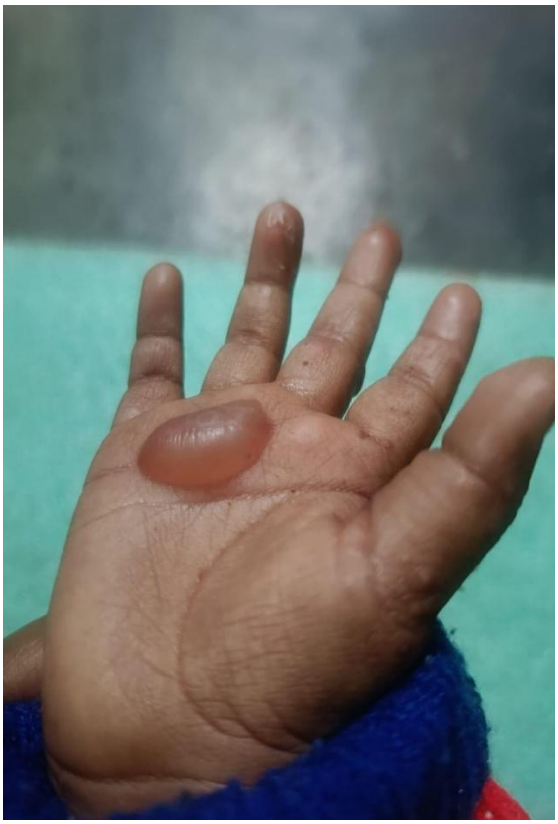

Figure 5

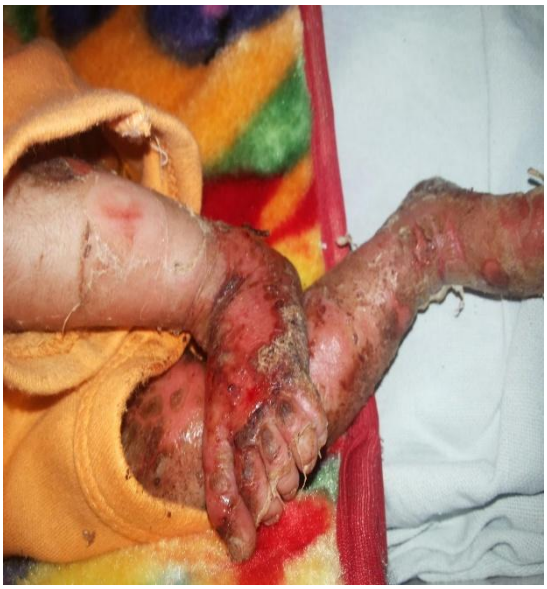

Figure 6

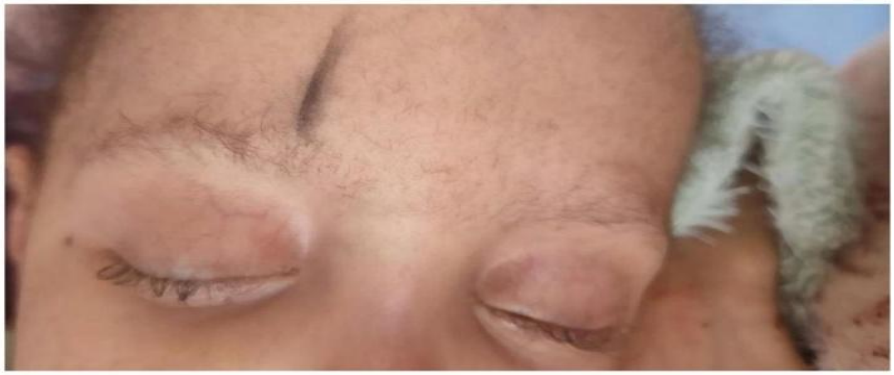

Figure 7

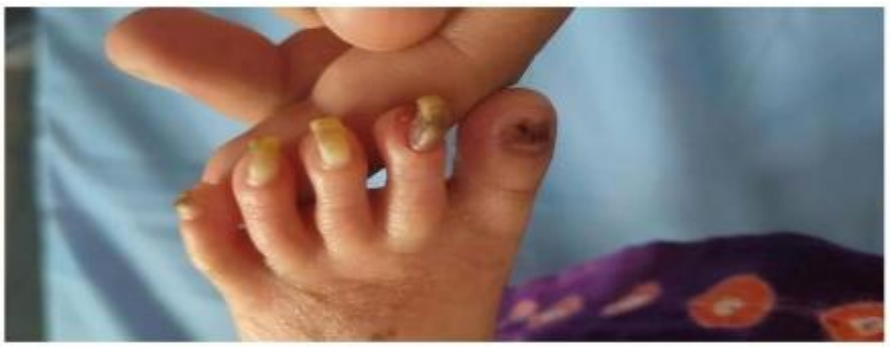

Figure 8

**DISCUSSION**

Genodermatoses refers to a group of disorders characterized by genetic abnormalities affecting the structure and function of the skin. It can have multisystem involvement. The spectrum of genodermatoses is huge with varied presentations in respect of the structure, function and morphology of the skin. The variation depends upon the ethnicity, race, religion, and geographical location. Our area of Rajouri has a mixed population with 62 percent muslims, 34 percent hindus and rest others.

In our study, we found a total of 52 patients over a period of 1 year. The prevalence in our study came out to be .29 whereas another study from Kashmir area reported a prevalence of .72 percent among a 97percent muslim majority area.[3] Another author confounded that that at least 1% of all livebirths had disorder inherited in a simple Mendelian fashion.[4]

Majority of the patients reported in the age group of 0-6 years in accordance with studies by authors who also reported increased prevalence in the first year of life.[5] The male to female ratio in our study was 1.2 similar to few other studies.[6] In our study a high degree of consanguinity was noted in 98% of the muslim patients. In 12 patients family history was present. Similar findings were observed in studies done over regions with almost similar topographical and ethnic populations.[3,7]

We did observe a varied spectrum of genodermatoses. Most common being the genetic disorders of keratinization with icthyosis being dominant. Similar results were observed in two other studies.[3,8]. However in our study we found cases of aquagenic syringeal keratoderma biopsy proved. Another rare case of Naxos syndrome was reported and genetically tested. No case of Naxos syndrome has earlier been reported from Jammu and Kashmir.

In our study, vascular malformations formed a major part at second position followed by familial tumor syndromes and DNA instability syndromes. Another study reported highest incidence of familial tumor syndrome among their patients.[9] Among the familial tumor syndromes and DNA instability disorders, most common was neurofibromatosis and tuberous sclerosis which was in concordance with the above study.7 We reported neurofibromas, CALMs and axillary freckling in 100 percent of the patients wheras various studies have found variable results.[3,10]

Among patients of tuberous sclerosis, angiofibromas and ash leaf macules were reported in majority but systemic complaints and various other features were rarely reported in our study. Various other studies have also reported very high incidences of angiofibromas.[11,12,13] But unlike our study they also reported various clinical and systemic complaints also. This could be attributed to varied age of presentations to the opd.

The next common group was of genetic blistering disorders. We did find a higher percentage of patients suffering from blistering disorders of genetic origin as compared to various other studies[5,6] where low prevalence was found barring one study from Kashmir with similar prevalence.1 most of the patients were of epidermolysis bullosa simplex where diagnoses was made clinically where as one patient was proven of to be ectodermal dysplasia-skin fragility syndrome(EDS-SFS) proven by genetic testing. We could not find any other case report of ectodermal dysplasia-skin fragility syndrome(EDS-SFS) form Jammu and Kashmir state.

Other patients were of genetic disorders of pigmentation. We found oculocutaneous albinism and dyschromatoses universalis hereditaria.

Xeroderma pigmentosa(XP) was clinically diagnosed in one patient and he gave history in another sibling. The patients had Basal cell carcinoma also which was biopsy proven. Early onset of skin cancers are proven in XP further aggravated by higher altitudes in our region. Miscellaneous disorders were also reported.

Genodermatoses is still a challenge in this era of technology in terms of diagnoses and management . things are further made difficult in resource poor settings where genetic testing and chromosomal mapping are not available. Thus clinical and histopathological diagnoses still remains the tool.

There is a dire need of more and more studies on geneodermatoses along with a database or registry. We need to inculcate the use of newer technologies like artificial intelligence etc.for easier and faster recognition of these and prevention at premarital level. More studies are needed as the genedermatoses is not only prevalent in our region but various studies from different parts of India and the world have reported a higher prevalence too.[14,15,16]

**Conclusion**

We feel that in the perspective of resource poor nations like India, we need to have more and more studies from various regions of our country on geneodermatoses which will surely facilitate the development of larges databases for early and accurate diagnoses. Moreso, this will improve awareness among the parents and patients as well for multicentric management options.

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