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Cover image: Destructive capillary hemangioma of childhood; multiple carcinomas of face and large melanomas on the scalp in xeroderma pigmentosum; giant congenital hairy nevus, see article on page 307.
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Medical genetics

The spectrum of genodermatoses and congenital cutaneous conditions in northern Ethiopia

Federica Dassoni¹, MD, Aldo Morrone², MD, and Valeska Padovese¹, MD

¹Italian Dermatological Center of Ayder Referral Hospital, Mekele, Ethiopia.
²S. Camillo Forlanini Hospital, Rome, Italy.
²National Institute for Health, Migration and Poverty (NIHMP), Rome, Italy

Correspondence
Federica Dassoni, MD
Italian Dermatological Center of Ayder Referral Hospital
Mekele
Ethiopia
E-mail: federica.da@gmail.com

Present address
Federica Dassoni, MD
Operative Unit of Dermatology, University of Milan, I.R.C.C.S. Foundation, Ca Granda Ospedale Maggiore Policlinico, via Pace, 9 20125, Milan, Italy

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Abstract
Reports of congenital diseases in Africa are scanty, probably because of their rarity, the lack of knowledge among health workers, and the difficult political and social situation in different African countries. We describe here the spectrum of genetic and rare congenital cutaneous conditions encountered at the Italian Dermatological Center of Ayder referral hospital of Mekele, Ethiopia, over a 3-year period. All patients attending the Italian Dermatological Center were registered in a database, and medical records of genetic and congenital disorders diagnosed from January 2008 to December 2010 were retrospectively analyzed. Over the total, 24 different genetic and congenital disorders affecting 122 individuals (0.4% of the total case load) were observed. In our case series, we did not report any patient affected by albinism, in contrast with literature from other African countries. To our knowledge, this is the first report from northern Ethiopia. A brief update on the commonest disorders is included.

Introduction
Reports of genetic and congenital skin diseases in Africa are scanty, probably because of their rarity but also due to challenges facing resource poor medical services, such as limited access to medical care and lack of health workers and patient awareness about these rare cutaneous conditions.

Aimhth., palmoplantar keratodermas (PPK), congenital ichthyoses, albinism, xeroderma pigmentosum (XP), neurofibromatosis (NF1), and incontinentia pigmenti achromians have been described in Africa.¹

Regarding genetic (or hereditary) disorders in the text, we mean diseases transferred from one or both parents to one or more of their children because of specific abnormalities (mutations) in their genes. Specifically, we refer to Mendelian disorders caused by mutations that occur in the DNA sequence of a single gene.

Regarding congenital conditions, we mean structural abnormalities of the skin that exist at, and usually before, birth regardless of their causation.

Our study was performed at the Italian Dermatological Center (IDC) of Mekele, the capital city of Tigray region, northern Ethiopia. IDC is the only referral and training center for skin diseases, diagnosis, and care in the region. A staff of Italian and local dermatologists treats an average of 70 patients per day coming from urban and rural districts.² The Tigray Regional State has a total land area of about 80,000 km² and an estimated population of 4,565,000 (July 2008).³ In our area, we observed that some genetic diseases are relatively well represented, such as XP, ichthyoses, NF1, and tuberous sclerosis (TS), but this has not been documented yet. To our knowledge, this is the first report from northern Ethiopia.

The aim of this study is to describe clinical characteristics of genetic and congenital cutaneous disorders presenting at the IDC, to raise awareness of their range in northern Ethiopia, and to highlight challenges facing dermatological services in diagnosing these rare conditions due to lack of facilities for diagnosis and treatment. The gathered data can be used as a rough
reflection of the presence of genetic and congenital cutaneous disorders in the region.

Materials and methods
This is a retrospective analysis of medical records. All patients attending IDC were examined by an Italian dermatologist. Diagnoses were registered in a database, and medical records of genetic disorders from January 2008 to December 2010 were analyzed. Diagnoses were clinical and, when possible, confirmed by skin biopsy. Owing to our limited medical facilities, gene mutations were not further investigated.

Results
Over the total, 24 different genetic and congenital cutaneous disorders affecting 122 individuals (0.4% of the total caseload) were observed in the 3-year period (Table 1). The following categories were particularly well represented.

Transient symptomatic zinc deficiency (18 cases) has not been previously described in Africa, and it is a relatively frequent disease in northern Ethiopia. It presents in breastfed infants with cutaneous manifestations indistinguishable from acrodermatitis enteropathica, which is autosomal recessive and requires lifelong zinc supplementation. Our diagnosis was based on clinical features and on the prompt and impressive response of just a few days of oral zinc supplementation. Our patients were mainly female (11 of 18). Symptomatic zinc deficiency in full-term infants is usually associated with heterozygosity for deletion of gene SLC39A4 and/or low zinc levels in mother’s milk.10 Dietary zinc deficiency in pregnant women could be an associated cause.11

Ichthyoses (autosomal recessive, autosomal dominant, and X-linked) were observed in 18 patients, three of which were infants presenting with colloidon baby features. Most of the patients were males (15 of 18). Most cases were X-linked ichthyosis and lamellar ichthyosis, with one case of keratinocthis ichthyosis and three not defined (probably ichthyosis vulgaris). The three cases of colloidon baby presented between two and four months of age, but the disorder was referred since birth, which reflects late access to health facilities from rural people.

NF1 is an autosomal dominant condition characterized by extremely variable expressivity. Café-au-lait (milk and coffee) spots and neurofibromas are the defining features.12 13 In dark-skinned people, café-au-lait spots appear darker than in European populations.

NF1 was observed in 15 patients. The male to female ratio was 2:1. Most patients were adults (mean age 31); this reflects the tendency of seeking medical advice only when major problems appear. Some complications were malignancy in one patient, epilepsy and spinal bone deformity in one, and tumors in three, and itching in four. One patient (M, 60) presented with clinical features of bilateral mosaic NF1 (Fig. 1). This presentation is very rare, less severe than classic NF1, and associated with a lower risk of transmission to the offspring. It may not be easy to differentiate the mosaic bilateral form and classic NF1, although it would be important to assess the risk for the offspring.14 15

Infantile capillary hemangioma was observed in 10 patients. This is a common condition both in Western and African countries. One patient presented with a large and destructive hemangioma of the face, with deformity of the nose and mouth (Fig. 2). A second had a lesion causing deformity and destruction of the left ear. Two patients presented with an eye localization causing closure of the eyelid. One child presented multiple small angiomas spread over the body, in the number of tens, without any systemic symptoms. Therapeutic options for infantile hemangiomas are limited in developing countries. Beta-blockers are sometimes available but rarely used by health workers.

XP was observed in seven patients. Two of them were siblings. XP is a rare genetic disorder characterized by defective DNA repair in cells damaged by ultraviolet light.13 Inheritance is autosomal recessive, and it is associated with a high incidence of consanguinity in parents, especially first cousins. XP is generally regarded as a very serious disease in the tropics because of its pronounced sensitivity to sunlight.14 Our patients presented different grades of disease severity, despite the common environment characterized by high sunlight.15 16

Photoprotection from birth is unlikely to occur as there are no facilities for prenatal diagnosis of XP and thus the diagnosis will not be made early. Only one of our patients was diagnosed before one year of age. For these patients, the prognosis is gloomy (Fig. 3).

Port-wine stains (flat angiomas) were observed in six patients. This is a sporadic presentation and most of the time harmless. Sometimes it can be associated with deep vascular malformations as in the Sturge-Weber syndrome or Klippel-Trenaunay and Parkes-Weber syndrome, which are rare. Only one of our patients presented with the clinical features of Sturge-Weber syndrome.17

Ocular and neurologic manifestations are due to deep choroidal or meningeal angiomatosis. Cerebral lesions are easily seen with computerized tomography or magnetic resonance imaging, although we did not have any access to these examinations.

Epidermolysis bullosa congenita was observed in five patients. From the clinical features, we classified one case of junctional type in a 1-month-old girl; one case of
<table>
<thead>
<tr>
<th>Disease, transmission</th>
<th>N</th>
<th>M/F (age)</th>
<th>Notes</th>
<th>Histology performed</th>
<th>Associations</th>
</tr>
</thead>
<tbody>
<tr>
<td>Symptomatic zinc deficiency (sporadic)</td>
<td>18</td>
<td>M/F - 7 : 11 (4-20 months)</td>
<td>1 patient with moderate malnutrition</td>
<td>No</td>
<td>1: unknown chromosome anomaly</td>
</tr>
<tr>
<td>Neurofibromatosis 1 (AD)</td>
<td>15</td>
<td>M/F - 10 : 5 (4-64 years)</td>
<td>1 patient with mosaic bilateral NF1</td>
<td>Yes, in 2 cases</td>
<td>1: malignancy</td>
</tr>
<tr>
<td>Ichthyosis</td>
<td></td>
<td>M - 15 (4-20 years)</td>
<td></td>
<td>No</td>
<td>1: epilepsy and bone deformity</td>
</tr>
<tr>
<td>Total</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Lamellar i. (AR, AD)</td>
<td>7</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>X-I</td>
<td>4</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Other/not classified</td>
<td>4</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Collodion baby</td>
<td>3</td>
<td>F = 3 (2-4 months)</td>
<td>1 patient from Atar region</td>
<td>No</td>
<td></td>
</tr>
<tr>
<td>Infantile capillary hemangioma</td>
<td>10</td>
<td>M/F (infants)</td>
<td>Causal deformity in 2 cases</td>
<td>No</td>
<td></td>
</tr>
<tr>
<td>Xeroderma pigmentosum (AR)</td>
<td>7</td>
<td>M/F = 4 : 3 (3-21 years)</td>
<td>1 patient from Atar region</td>
<td>Yes, in 4 cases</td>
<td>3 developing MM, all developing NMSC</td>
</tr>
<tr>
<td>EBC</td>
<td></td>
<td>M/F = 4 : 1 (4 months-6 years)</td>
<td>2 brothers with epidermolytic</td>
<td>No</td>
<td>Dystrophy of fingers/toes</td>
</tr>
<tr>
<td>Total</td>
<td>5</td>
<td></td>
<td>EBC Dowling Meara type:</td>
<td></td>
<td></td>
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<tr>
<td>Dermolytic (AD, AR)</td>
<td>1</td>
<td></td>
<td>3 from Atar region</td>
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<td></td>
</tr>
<tr>
<td>Junctional (AR)</td>
<td>1</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Epidermolytic (AD, sporadic)</td>
<td>3</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Palmoplantar keratodermia</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Total</td>
<td>5</td>
<td>M/F = 4 : 1 (4-46 years)</td>
<td></td>
<td>No</td>
<td>1: epilepsy</td>
</tr>
<tr>
<td>Grether disease (AD)</td>
<td>1</td>
<td>M (23 years)</td>
<td></td>
<td></td>
<td>(Sturge–Weber syndrome)</td>
</tr>
<tr>
<td>Papillon–Leberre syndrome (AR)</td>
<td>2</td>
<td>F (3 years); M (25 years)</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Urna–Thost disease (AD)</td>
<td>1</td>
<td>M (4 years)</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Unknown</td>
<td>1</td>
<td>M (27 years)</td>
<td></td>
<td>No</td>
<td></td>
</tr>
<tr>
<td>Flat angioma (sporadic)</td>
<td>6</td>
<td>M/F = 4 : 2 (4-46 years)</td>
<td></td>
<td>No</td>
<td></td>
</tr>
<tr>
<td>Sturge–Weber syndrome</td>
<td>1</td>
<td>F (12 years)</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Blashkoid nevus (mosaic, sporadic)</td>
<td>5</td>
<td>F = 4, M = 1 (2-25 years)</td>
<td></td>
<td>No</td>
<td></td>
</tr>
<tr>
<td>Tuberous sclerosis (AD)</td>
<td>5</td>
<td>F = 3, M = 2 (6-28 years)</td>
<td>1: mosaic tuberous sclerosis</td>
<td>No</td>
<td></td>
</tr>
<tr>
<td>Verrucom epidermolytic nevus (mosaic, sporadic)</td>
<td>4</td>
<td>F = 3, M = 1 (8-10 years)</td>
<td>1: father presented localized lesion</td>
<td>Yes, in 2 cases</td>
<td></td>
</tr>
<tr>
<td>Segmental hypomelanosis (lto) (mosaic, sporadic)</td>
<td>4</td>
<td>M = 4 (4-42 years)</td>
<td></td>
<td>No</td>
<td></td>
</tr>
<tr>
<td>Segmental speckled nevus (sporadic)</td>
<td>4</td>
<td>F = 4 (18-32 years)</td>
<td></td>
<td>No</td>
<td></td>
</tr>
<tr>
<td>Cavernous hemangioma (sporadic)</td>
<td>3</td>
<td>M = 2, F = 1 (6-53 years)</td>
<td></td>
<td>No</td>
<td></td>
</tr>
<tr>
<td>Darier's disease (AD)</td>
<td>2</td>
<td>F = 2 (18 and 23 years)</td>
<td>None with neurologic symptoms</td>
<td>Yes, in 2 cases</td>
<td></td>
</tr>
<tr>
<td>Porphyria cutanea tarda</td>
<td>2</td>
<td>M = 2 (34 and 57 years)</td>
<td></td>
<td>No</td>
<td></td>
</tr>
<tr>
<td>Giant congenital nevus (sporadic)</td>
<td>1</td>
<td>F (3 years)</td>
<td></td>
<td>No</td>
<td></td>
</tr>
<tr>
<td>Neithers- Dantas syndrome (AD, AR, XL)</td>
<td>1</td>
<td>M (19 years)</td>
<td></td>
<td>No</td>
<td></td>
</tr>
<tr>
<td>Mastocytosis (sporadic)</td>
<td>1</td>
<td>M (4 months)</td>
<td></td>
<td>No</td>
<td></td>
</tr>
<tr>
<td>Peaboldism</td>
<td>1</td>
<td>F (5 months)</td>
<td></td>
<td>No</td>
<td></td>
</tr>
<tr>
<td>Peutz–Jeghers syndrome (AD)</td>
<td>1</td>
<td>F (6 years)</td>
<td></td>
<td>No</td>
<td></td>
</tr>
<tr>
<td>Chromosome anomalies</td>
<td>2</td>
<td>M = 2</td>
<td>Unknown, presenting with atypical facies (hypertelorism, finger anomalies, etc.)</td>
<td>No</td>
<td></td>
</tr>
</tbody>
</table>
Table 1 Continued

<table>
<thead>
<tr>
<th>Disease, transmission</th>
<th>N</th>
<th>M/F (age)</th>
<th>Notes</th>
<th>Histology performed</th>
<th>Associations</th>
</tr>
</thead>
<tbody>
<tr>
<td>Metabolic anomalies</td>
<td>2</td>
<td>M = 1, F = 1</td>
<td>Unknown, presenting failure to thrive, irritability, skin alterations, etc.</td>
<td>No</td>
<td></td>
</tr>
</tbody>
</table>

Total: 122

AD, autosomal dominant; AR, autosomal recessive; EBC, epidermolytic bullosa congenita; MM, malignant melanoma; NMSC, non-melanoma skin cancer; XL, X-linked.

![Figure 1](image1.png) Mosaic bilateral neurofibromatosis with partial unilateral lentiginosis

![Figure 2](image2.png) Disfiguring capillary hemangioma of childhood

...retardation. TS may be autosomal dominant or, more frequently, due to spontaneous mutation. Our patients did not have a family history of the disease. Only one of them had history of seizures, none had evidence of mental retardation. One presented with clinical features of mosaic TS, with unilateral angiofibromas on the left side of the face and forehead. Few reports from African countries are found.

PPK are more common in Africa than in Europe but are difficult to classify, and their hereditary nature is not always recognized.” The most common forms are dominantly inherited with a relatively mild presentation. The rarer forms are mainly recessively inherited, have more widespread lesions, and are often associated with abnormalities of eyes, teeth, hair, and/or cerebral function. In the hereditary keratodermas, involvement is usually bilateral and symmetrical. We observed five cases of PPK,...
classified clinically as Greither’s disease (autosomal dominant; one case), Papillon-Lefèvre syndrome (autosomal recessive; two), Unna-Thost disease (autosomal dominant or sporadic; one), and not classified (one). The latter presented with non-transgradient hyperkeratosis and deformity of the toes, fingers, and nails.

In Ethiopia, Papillon-Lefèvre syndrome has never been reported previously.17–18 Verrucous epidermal nevi (non-epidermylic) are congenital, non-inflammatory cutaneous hamartomas composed of keratinocytes. They are probably heterogeneous, representing mosaicism for different mutations. Their general prevalence in adults is probably 0.1–0.5%.29 Prevalence in northern Ethiopia is not known. We observed five patients with simple verrucous nevi disposed along Blaschko lines, indicating an early mutation of a clone of cells during embryogenesis, with a mosaic pattern.

Verrucous epidermylic nevi was observed in four patients and confirmed by skin biopsy, which showed histological features of epidermylic hyperkeratosis. Patients with epidermylic epidermal nevi can give rise to children with a rare but serious condition known as congenital bullous ichthyosiform erythroderma. This can happen when the mutation involves the gonads. Keratin mutations have been described in both conditions, and the relationship between the two can be explained using the concept of genetic mosaicism.30–32 Our patients presented with different degrees of body surface area involvement. Verrucous lesions were disposed along the lines of Blaschko either unilaterally or bilaterally. Discriminating between the above and segmental verrucous nevus and lichen striatus (which is transitional) may be challenging without histology.

Hypomelanosis of Ito (previously also called incontinence pigmenti achromians) is a mosaic disease associated in part with a chromosomal alteration. It is usually a sporadic disorder.33–35

The cutaneous lesions are associated in three-quarters of patients with mental retardation and seizures. Skin lesions consist of hypopigmented macules arranged in band-like streaks and whorls along Blaschko lines. The bands may be unilateral or bilateral and resemble systematized nevus depigmentosus, to which it may be related.36,37 We observed four cases with the typical dermatological features of hypomelanosis of Ito. Not one of our patients had evidence of associated neurologic symptoms.

Darier’s disease (DD) is a rare genetic disease with an autosomal dominant, variable penetrating transmission. It is characterized by abnormal keratinization in the epidermis, nails, and mucosa. DD can present as a generalized autosomal dominant condition as well as a localized or segmental postzygotic condition. Hypopigmented guttate macules (1–5 mm sized) have also been described as a rare feature typical of dark-skinned individuals.38 DD has been described in sub-Saharan Africa but not in Ethiopia.39–40 We observed two patients with DD. They had diffuse cutaneous manifestations, one of them presenting with associated hypopigmented guttate macules. Both patients were late presentations and had significant social problems and impaired life quality mainly due to the malodorous complications of their dermatosis.

Giant congenital melanocytic nevi are benign proliferations of cutaneous melanocytes apparent at birth or in the first few weeks thereafter. They can be recognized not only by their size (>20 cm) but also by their ability to affect deep dermal layers and other subcutaneous tissues.41

Neurocutaneous melanosis is characterized by an increased number of melanocytes and melanin deposit in the central nervous system associated with giant melanocytic congenital nevi. In most patients, the neurocutaneous melanosis is asymptomatic, only detectable by magnetic resonance imaging; those patients with clinical manifestations have a poor prognosis.42 We observed one child with giant congenital melanocytic nevus, diffuse on the back, neck, shoulders, and legs, with multiple satellite lesions. Lesions were hairy and, on the back, subcutaneous nodules were present (Fig. 4). No signs of neurologic involvement were clinically evident.

Few reports from Africa are found.43–44

Ehlers-Danlos syndrome (EDS) is a heterogeneous group of connective tissue disorders characterized by
hyperextensibility, joint hypermobility, skin fragility, cigarette-paper scarring over bony prominences, mitral valve prolapse, and other findings (e.g., osteoporosis, congenital hip dislocation, motor delay, etc.). At least 10 different subtypes of EDS have been classified based on genetic, biochemical, and clinical characteristics. They may be transmitted in an autosomal dominant, autosomal recessive, or X-linked way. We observed a 19-year-old boy with atrophic (cigarette paper) scars on knees and elbows, who could not stand up and walk. He was previously examined by several doctors and had x-ray examinations of lower limbs, but he never had a diagnosis. The skin was doughy/velvety to touch and deformable. Bruising was difficult to examine because of the dark skin color. Clinically, the most likely diagnosis was EDS type II. This is to our knowledge the first case report from Ethiopia. Two cases only are reported from sub-Saharan Africa, namely from Zimbabwe and South Africa.

Peutz-Jeghers syndrome is an inherited autosomal dominant disorder characterized by peri-orificial mucocutaneous pigmentation and gastrointestinal polyps. Peutz-Jeghers syndrome also assumes more importance because of the higher risk of intestinal and extra-intestinal cancers. We observed one child with peri-orificial lentiginosis. Our diagnosis was only clinical and could not be confirmed with endoscopy. Few reports in African patients are found, from Ethiopia and Nigeria.

**Discussion**

Genetic and congenital cutaneous conditions are rare worldwide. In Africa, as in any resource-poor setting, they may remain undiagnosed or misdiagnosed due to poor knowledge of primary health workers, lack of dermatologists, and access barriers to referral hospitals. Thus, the epidemiological distribution of these conditions in many African countries is only partially known.

In contrast with the literature, which reports only sporadic cases of genetic and congenital cutaneous conditions from sub-Saharan Africa, we observed a number of genetic and congenital skin diseases in northern Ethiopia, some of which were rarely, if ever, described in other African countries (e.g., symptomatic zinc deficiency, EDS, epidermolysis bullosa congenital, TS, porphyria). Surprisingly, in Tigray we did not observe any patient affected by albinism, which is well described in other countries. This should encourage us to further investigate genodermatoses distribution and prevalence in the region, which could be different from other areas.

This is to our knowledge the first report of genetic and congenital dermatoses from northern Ethiopia. Health education addressed both to health personnel and to families, social support, and funding would help to improve health workers’ diagnostic and counseling capabilities, increase social acceptance, and strengthen the reporting system.

As sophisticated diagnostic techniques such as computed tomography scan, heart ultrasound, blood levels of chemicals, chromosome mapping, or loci identification are not available in resource-poor settings, we highlight the importance of clinical diagnosis, which can be confirmed in some cases by skin biopsy, to enable reasonably accurate diagnoses and support the patients and their families with prognostic information and simple therapeutic interventions.

Epidemiological data are not yet available; consequently, the burden of diseases could not be appreciated precisely. While the high numbers might represent selection bias due to the absence of other dermatologic centers in the region, another explanation could be consanguineous marriage, which is more frequent in our rural situation. A specific example of this so-called geographic endogamy is shown by the Afar nomad population, who live in the lowlands of Tigray region and of the neighboring Afar region.

We made a number of our diagnoses incidentally on patients attending a hospital for other reasons. Thus, it is difficult to make a correct estimation of their number. As our unit gains a reputation both with local colleagues and with the patients, we anticipate seeing a more representative presentation rate of these rare disorders.
The overriding challenge to the genetic or congenital skin disease patient in northern Ethiopia is that of stigma. Although this occurs elsewhere in the world, it is especially critical in African countries. This leads to exclusion from many aspects of life, such as school, religious gatherings, employment, and even family support. Long-term therapies are also difficult to perform because of the cost, lack of medications, and low compliance of patients that often use herbal remedies or consult traditional healers.

Premarital genetic counseling would be helpful for all couples with a genetic disease history in the family and in case of consanguinity. Unfortunately, formal premarital genetic counseling is not provided in the region.

In conclusion, we assessed for the first time genetic and rare congenital dermatoses in northern Ethiopia. In spite of the challenges facing care in this resource-poor setting, we feel there is value in accurate clinical diagnosis and prognosis. In time, by increasing our understanding of the epidemiology and disease burden, in the context of impact on quality of life, we hopefully will be able to manage health resources more effectively.

References


