7. What is Genetic counselling?

- Genetic counselling is a process of communication and education that addresses the concerns of couples with family history of Genodermatoses by a specialist in this field with thorough knowledge of the disease.
- Counselling regarding the mode of inheritance of the disease, possibility of children being affected, avoidance of consanguineous marriages (marriage between blood relatives), grading of severity of disorder in subsequent generations is done.

8. How can I manage the stress?

- Understand the disorder correctly with professional help.
- Have realistic expectations from the treatment offered.
- Take genetic counselling.
- Professional advice from a psychiatrist may be needed to cope with the stress.
- Joining in the self-help groups of genetic disorders to share the knowledge and empathize may help to relieve the stress.

9. What can I do to support the ongoing research projects?

- Proper maintenance of records and maintaining a central clinical registry will go a long way in helping the ongoing research in the field of Genodermatoses.
- The lack of awareness and rarity of these conditions are major obstacles in the management and the planning of research in this specialty.
- This is especially relevant in the context of our country where multi-specialty networking or multi-centre collaboration may not be feasible.
- The pertinent issue is how to tackle a problem where the magnitude is undefined.
- The first step, therefore, is setting up disease-specific national registry with detailed phenotypic inputs for confirmation of accurate diagnosis.

10. Are there support groups and self-help groups for these diseases in our country?

- Patient and physician help groups are coming up in India, gradually as the awareness about various Genodermatoses is spreading.

List of support groups and self-help groups:

1. Multi-disciplinary clinic and services available for the care of Epidermolysis Bullosa patients in Centre for Human Genetics, Bangalore (http://www.chg.res.in/)
2. Tuberous sclerosis India Support Group (http://www.tsicindia.org/)
3. Epidermolysis Bullosa without borders (http://www.debra-international.org)
4. The Neurofibromatosis Association (http://www.nfauk.org)
6. Organization of rare diseases in India (http://ordindia.org/)
7. Indian Genetic Disease Database (IGDD) (http://www.igdd.kcb.res.in)
8. Metabolic Errors and Rare Diseases organization of India (http://www.merdindia.com/)
10. Together Against Genodermatoses (http://www.tag-eu.org/)

Disclaimer:
This leaflet is only for general patient information and is not intended for self-medication. There is no legal liability of IADVL arising out of any adverse consequences to the patient subsequent to its use for self-treatment of the disease. Images are just for the depiction of the condition and are not to be used for any other purpose.

Website: Patient Information Leaflet
www.iadvl.org/patient-information-leaflet

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1. What are Genodermatoses?

Genodermatoses are rare inherited genetic diseases with skin changes.

The individuals who are at a higher risk are those children who are born with a family history of known Genodermatoses or a sibling affected with a genetic skin disease.

2. How does it present?

Genodermatoses are multi-system disorders, so apart from skin; other body systems are also affected.

Children can be symptomatic at birth or early childhood with specific and extensive skin changes. The skin may be dry, red and scaly and may have disease specific changes.

3. How can one prevent unborn siblings from getting affected?

It is important to be aware about the mode of inheritance of the Genodermatoses and to inform your Gynecologist about the condition in the family.

Timely pre-natal diagnosis is the key in preventing the unborn siblings' from getting affected.

List of few common genodermatoses (with skin manifestations)

- Ichthyosis and related disorders
- Neurofibromatosis
- Tuberous sclerosis
- Epidermolysis Bullosa
- Palmoplantar keratoderma
- Xeroderma pigmentosum
- Photosensitive Genodermatosis
- Incontinentia pigmenti
- Ectodermal dysplasias
- Epidermal nevi and related syndromes
- Vascular disorders
- Hypomelanosis of Ito and miscellaneous disorders

4. How can the parents take care of their baby who is suffering from a Genodermatoses?

As the parents take their baby home, they should be advised on these issues:

- Children with Genodermatoses require continued specialized care to identify early deviation of growth, development and behavior from normal. Hence, regular follow up in a specialized clinic is a must.

5. Are there any tests to confirm the diagnosis of Genodermatoses?

- Genodermatoses are rare and there are limited specialized centers in our country performing genetic studies. (Refer Annexure Centres for Genetic Testing)
- Dermatologists and pediatricians can make an initial diagnosis based on the clinical spectrum.

6. Is there any definitive therapy for Genodermatoses?

- There are several research projects going on across the world and mapping of the human genome has helped us in understanding the Genodermatoses and their mode of inheritance.
- Gene therapy studies are nearing completion and are a ray of hope for definitive therapy for some of the Genodermatoses.
- However with regular follow-up and supportive and symptomatic therapy your child can also lead a near normal life.