

## Basic knowledge about Genodermatoses

Provide basic information packages and links to useful websites to help find out more about Genodermatoses.

## Medical expertise

Regular updates and CME's can help in exchange of global information and advances in the field of Genodermatoses which can be passed on to the patients.

## Fundraising

With the spread of information various pharmaceutical and corporate houses can be approached for funds which can be utilized for providing better facilities to the patients suffering with these ailments.

## Campaigning and Awareness

To spread awareness about Genodermatoses and availability of prenatal diagnostic facilities.

## Infrastructure and networks

### Setting up a disease-specific national registry

- 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.

### Setting up a national Genodermatoses support group

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

- These support and self-help groups are promoting awareness about various genodermatoses and provide moral and disease informative support to the parents of such children.
- Campaigning about newborn screening is another important thrust area of these groups.

### 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.tscindia.org/>)
3. Epidermolysis Bullosa without borders (<http://www.debra-international.org/>)
4. The Neurofibromatosis Association (<http://www.nfauk.org/>)
5. Ichthyosis Support Group(National health portal: <http://www.nhp.gov.in/disease-a-z/i/ichthyosis> & <http://www.ichthyosis.org.uk/>)
6. Organization of rare diseases in India (<http://ordindia.org/>)
7. Indian Genetic Disease Database (IGDD) (<http://www.igdd.iicb.res.in>)
8. Metabolic Errors and Rare Diseases organization of India (<http://www.merindia.com/>)
9. List of International laboratories for Gene Tests (<http://www.ncbi.nlm.nih.gov/sites/genetests/>) and the Genetic Testing Registry (<http://www.ncbi.nlm.nih.gov/gtr/>)
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.

Weblink to Patient Information Leaflet:  
[www.iadvl.org / patient information leaflet](http://www.iadvl.org/patient-information-leaflet)

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## UNDERSTANDING GENODERMATOSES

## INDIAN ASSOCIATION OF DERMATOLOGISTS, VENEREOLOGISTS AND LEPROLOGISTS

- What are Genodermatoses?
- What happens in Genodermatoses?
- How does Genodermatoses impact everyday life?
- Which are commonly encountered Genodermatoses?
- Is there a cure for Genodermatoses?
- Why is it difficult to manage a child with Genodermatoses?
- Is there a ray of hope for patients with Genodermatoses?
- How can we make a difference in the field of Genodermatoses?

## 1. What are Genodermatoses?

- Genodermatoses are a large group of inherited disorders with skin manifestations. Many of these disorders are rare.

## 2. What happens in Genodermatoses?

- These are inherited multisystem disorders and apart from skin other internal organs are also affected. They are rare and can sometimes be life-threatening.
- Symptoms usually appear at birth or early in life and may severely affect children.
- They have a high impact on the quality of life of the patients and of their family and on the society: social exclusion, disability, short life expectancy.

## 3. How does Genodermatoses impact everyday life?

- The burden of severe Genodermatoses is huge for the patients and their family.
- Social exclusion, disability, short life expectancy makes this population very vulnerable.
- Access to school, to work, and to leisure activities is often very difficult for the affected children.
- Frequent medical follow ups are required and in most of the cases it is only symptomatic therapy which is available as on date.
- There is also a risk that the other born and un-born siblings may have the same disease.

## 4. Which are commonly encountered Genodermatoses?

- Neurofibromatosis
- Tuberous sclerosis
- Ichthyosis and related disorders
- Epidermolysis Bullosa
- Palmoplantarkeratoderma
- Xerodermapigmentosum

- Photosensitive Genodermatosis
- Incontinentiapiigmenti
- Ectodermal dysplasias
- Epidermal nevi and related syndromes
- Vascular disorders
- Hypomelanosis of Ito and miscellaneous disorders

## 5. Is there a cure for Genodermatoses?

- Only symptomatic treatments are available in most of the diseases and they are of major importance.
- The medical care involves many specialties: dermatology, otolaryngology, pediatrics, gynecology, urology, ophthalmology, oncology, plastic and maxillofacial surgery, gastroenterology, nephrology, radiology, radiotherapy, odontology, nutrition, etc.

## 6. Why is it difficult to manage a child with Genodermatoses?

- The rarity of the conditions and lack of awareness are major obstacles in the management and the planning of research in this specialty.
- Multiple organ systems may be affected and treatment is mostly symptomatic with no definitive cure.

## 7. Is there a ray of hope for patients with Genodermatoses?

- There is now better understanding of the genetic basis of Genodermatoses with tremendous progress in their molecular diagnosis.
- The knowledge of genetic mutations facilitates the determination of gene function and correlation of the genotype with phenotype. The latter is especially relevant for prognosis and to make informed management decisions.
- Mutation analysis permits the offering of appropriate genetic counseling and makes DNA-based prenatal diagnosis feasible in high risk families.
- With the completion of the human gene project in 2003, we now have a mutation and single nucleotide polymorphism database of the majority of single gene inherited skin disorders.

- Perhaps the greatest advantage of the advances made in genotyping is the development of somatic gene therapy; for example, gene replacement therapy for autosomal recessive epidermolysis bullosa.

- It is imperative to remember that a lot still needs to be done; the genes responsible for some Genodermatoses remain unidentified and the pathomechanisms of some needs elucidation.

- Several Genodermatoses have a multi-system involvement resulting in severe morbidity and mortality that necessitates more focus on this specialty.

- Each country should have a national database facilitating screening, diagnosis, and management of patients.

- The professional physician bodies should encourage formation of self-help groups to cope with debilitating disease and improve the quality of life of the patients.

- Greater collaboration should be initiated between centers to recruit a larger cohort of patients and obtain more meaningful results.

- The use of alternative methods of diagnosis may also be warranted.

- Lastly, Genodermatoses are amenable to teledermatology and international collaboration with well established international laboratories.

## 8. How can we make a difference in the field of Genodermatoses?

### Making contacts

Asking patients form self- help groups which can further co-ordinate and contact similar groups in various regions.

### Capacity building

#### Advocacy

Encourage patients to find and join with others with Genodermatoses to form a strong patient-based support group. By strengthening the member base of families living with Genodermatoses, they will have a better chance to change government policies, as well as raise awareness and funds for Genodermatoses.