

## 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.

- 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/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.merdindia.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:

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# GENODERMATOSES

## INDIAN ASSOCIATION OF DERMATOLOGISTS, VENEREOLOGISTS AND LEPROLOGISTS

- What are Genodermatoses?
- How does it present?
- How can one prevent unborn siblings from getting affected?
- How can the parents take care of their baby who is suffering from a Genodermatoses?
- Are there any tests to confirm the diagnosis of Genodermatoses?
- Is there any definitive therapy for Genodermatoses?
- What is Genetic counselling?
- How can I manage the stress?
- What can I do to support the ongoing research projects?
- Are there Support groups and self-help groups for these diseases in our country?



## 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 family history of known Genodermatoses or a sibling affected with a genetic skin disease.



### 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

## 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.



## 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.

- Please bring your follow up record book which has details of the problems and management given to your child.
- Your baby may require visit to multiple specialists for overall assessment namely neonatologist, developmental pediatrician and therapist, dermatologist, ophthalmologist, audiologist & speech therapist, physiotherapist & occupational therapist, geneticist, pediatric and orthopedic surgeon.
- Follow-up at least twice a year is mandatory. Issues to be addressed during follow up are:
  - Monitoring weight, length and head circumference measurement
  - Dermatological assessment
  - Neurological and neuro-behavioral examination
  - Development assessment
  - Ophthalmological assessment
  - Hearing, Language and speech
  - Radiological assessment- Ultrasound head or MRI Brain
  - Feeding and supplementation
  - Immunization
  - Cognitive and intellectual assessment
- **Bathing:** Bathe your baby daily and use luke-warm water. Avoid using strong soaps and disinfectants. Do not scrub the skin frequently.
- **Clothing:** Always wrap your baby in multiple layers (one layer more than what you would require), the first layer should be soft cotton cloth. Always cover head, hands and feet with mitten and socks.
- **Eye care:** Clean with swab soaked in sterile water (boiled and cooled) from inside to outside using different swab for each eye. DO NOT APPLY KAJAL IN THE EYES.
- **Oil Massage:** Can be done with a non-irritant oil (any oil which is available eg. plain coconut oil, olive oil). Observe for any rashes.

## 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.