सत्यमेव जयते

कर्मचारी राज्य बीमा निगम
Employees' State Insurance Corporation

OPERATIONAL GUIDELINES for RARE DISEASES

INDEX

INTRODUCTION

The field of rare disease is complex & heterogeneous. New rare diseases and conditions are being identified and getting reported regularly in medical literature. Apart from a few rare diseases, the field is still at a nascent stage thereby posing difficulty in development of a comprehensive policy.

WHO defines rare disease as often debilitating lifelong disease or disorder condition with a prevalence of 1 or less, per 1000 population. Different countries have their own definitions to suit their specific requirements and suitable in context to their own population, health care system and resources.

Diagnosis of rare diseases is a challenge owing to lack of awareness in general public as well as in the medical profession. Effective and safe treatment is not available for most of the rare diseases even after diagnosis. About 95% of rare diseases have no treatment & less than one in ten patients receive disease specific treatment. Wherever drugs are available, they are prohibitively expensive, placing immense strain on resources.

In the absence of epidemiological data and data on how many patients suffer from diseases considered as rare in other countries understanding of the extent of burden, their associated mortality, morbidity and economic burden in India is impeded. Indian experts have identified and categorized rare diseases in three groups based on their clinical experiences. Till date only 450 diseases have been recorded in India.

Following group of disorders are identified and categorized by experts:

Group 1: A. Disorders amenable to one-time curative treatment: a) Disorders amenable to treatment with Hematopoietic Stem Cell Transplantation (HSCT) –

1 Such Lysosomal Storage Disorders (LSDs) for which Enzyme replacement Therapy (ERT) is presently not available and severe form of Mucopolysaccharoidosis (MPS) type I within first 2 years of age.

2 Adrenoleukodystrophy (early stages), before the onset of hard neurological signs.

3 Immune deficiency disorders like Severe Combined Immunodeficiency (SCID), Chronic Granulomatous disease, Wiskot Aldrich Syndrome, etc.

4 Osteopetrosis

5 Fanconi Anemia

6 Others if any to be decided on case to case basis by a technical committee.

B. Disorders amenable to organ transplantation

1 **Liver Transplantation** -Metabolic Liver diseases:

1 Tyrosinemia

2 Glycogen storage disorders (GSD) I, III and IV due to poor metabolic control, multiple liver adenomas, or high risk for Hepatocellualr carcinoma or evidence of substantial cirrhosis or liver dysfunction or progressive liver failure

3 MSUD (Maple Syrup Urine Disease)

4 Urea cycle disorders

5 Organic acidemias

2 Renal Transplantation

1 Fabry's disease

2 Autosomal recessive Polycystic Kidney Disease (ARPKD)

3 Autosomal dominant Polycystic Kidney Disease (ADPKD) etc.

4 Patients requiring combined liver and kidney transplants can also be considered if the same ceiling of funds is maintained. (Rarely Methyl Malonic aciduria may require combined liver & Kidney transplant) etc.

Group 2: Diseases requiring long term / lifelong treatment having relatively lower cost of treatment and benefit has been documented in literature and annual or more frequent surveillance is required:

A Disorders managed with special dietary formulae or Food for special medical purposes (FSMP)

1 Phenylketonuria (PKU)

2 Non-PKU hyperphenylalaninemia conditions

3 Maple Syrup Urine Disease (MSUD)

4 Tyrosinemia type 1 and 2

5 Homocystinuria

6 Urea Cycle Enzyme defects

7 Glutaric Aciduria type 1 and 2

8 Methyl Malonic Acidemia

1 Propionic Acidemia

2 Isovaleric Acidemia

3 Leucine sensitive hypoglycemia

9 Galactosemia

1 Glucose galactose malabsorbtion

2 Severe Food protein allergy

B. Disorders that are amenable to other forms of therapy (hormone/specific drugs)

1 NTBC for Tyrosinemia Type 1 13

2 Osteogenesis Imperfecta – Bisphosphonates therapy

3 Growth Hormone therapy for proven GH deficiency, Prader Willi Syndrome and Turner syndrome, others (to be decided on case to case basis by technical committee)

4 Cystic Fibrosis- Pancreatic enzyme supplement

5 Primary Immune deficiency disorders -Intravenous immunoglobulin therapy (IVIG) replacement eg. X-linked agammablobulinemia etc.

6 Sodium Benzoate, arginine, citrulline, phenylacetate (Urea Cycle disorders), carbaglu, Megavitamin therapy (Organic acidemias, mitochondrial disorders)

7 Others - Hemin (Panhematin) for Acute intermittent Porphyria, High dose Hydroxocobalamin injections (30mg/ml formulation – not available in India and hence expensive if imported)

8 Others (if any) to be decided on case-to-case basis, by a technical committee.

Group 3: Diseases for which definitive treatment is available but challenges are to make optimal patient selection for benefit, very high cost and lifelong therapy.

A Based on the literature sufficient evidence for good long-term outcomes exists for the following disorders

1 Gaucher Disease (Type I & III {without significant neurological impairment})

2 Hurler Syndrome [Mucopolysaccharisosis (MPS) Type I] (attenuated forms)

3 Hunter syndrome (MPS II) (attenuated form)

4 Pompe Disease diagnosed early (Both infantile & late onset)

5 Fabry Disease diagnosed before significant end organ damage

6 Spinal Muscular Atrophy

7 MPS IVA

8 MPS VI

B For the following disorders for which the cost of treatment is very high and either long term follow up literature is awaited or has been done on small number of patients

1 Wolman Disease

2 Hypophosphatasia

3 Neuronal ceroid lipofuscinosis

4 Cystic Fibrosis

5 Duchenne Muscular Dystrophy

As treatment modalities are few may not lead to "cure', exorbitantly costly and are not universally available & accessible, prevention needs to be the focus for all genetic disorders. Preventive strategies start with the education and awareness of general public and health care professionals. The prevention of genetic diseases is based on integrated and comprehensive strategy incorporating primary, secondary and tertiary level of prevention. Optimal screening and diagnostic strategy are recommended for those pregnant woman/newborn where there is a history of a child born with a rare disease and the diagnosis is confirmed.

Presently, the patients or beneficiaries of ESI suffering from Rare Diseases are referred for screening, diagnosis and treatment either to premier institutes or empanelled hospitals.

For diseases in group I and II, as the treatment is usually one time, rates are mostly available in CGHS rate list, cost involved in the treatment is lower than group III and is convenient to ESI beneficiaries, ESI is rendering treatment as per the existing system i.e. complete treatment for procedures which are available in CGHS rate list/drugs available in DG, ESIC RC without any capping of expenditure of Rs. 10 lakh per year per ESI patient. For treatment of non-CGHS procedure or drug not listed in DG, ESIC-RC, where expected expenditure is more than 10 lakh per year per ESI patient (especially for Group III diseases), the treatment is being provided after recommendation of the High Cost Treatment Committee and approval of the Hon'ble Chairman, ESIC.

For group III diseases currently, the above guidelines are being followed on case to case basis.

In order to deal with rare diseases ESIC Hqrs. has:

A Compiled a booklet "Operational Guidelines for Rare Diseases" (32 pages) for personnel involved in providing health care to ESI beneficiaries and encloses all the guidelines pertaining to diagnosis, screening, treatment, training and public awareness which includes:

   1 Organizational Structures for: Overall Management, Training, Data Collection

   2 Protocols for: Optimal Screening and Diagnosis, Management of confirmed or suspected cases

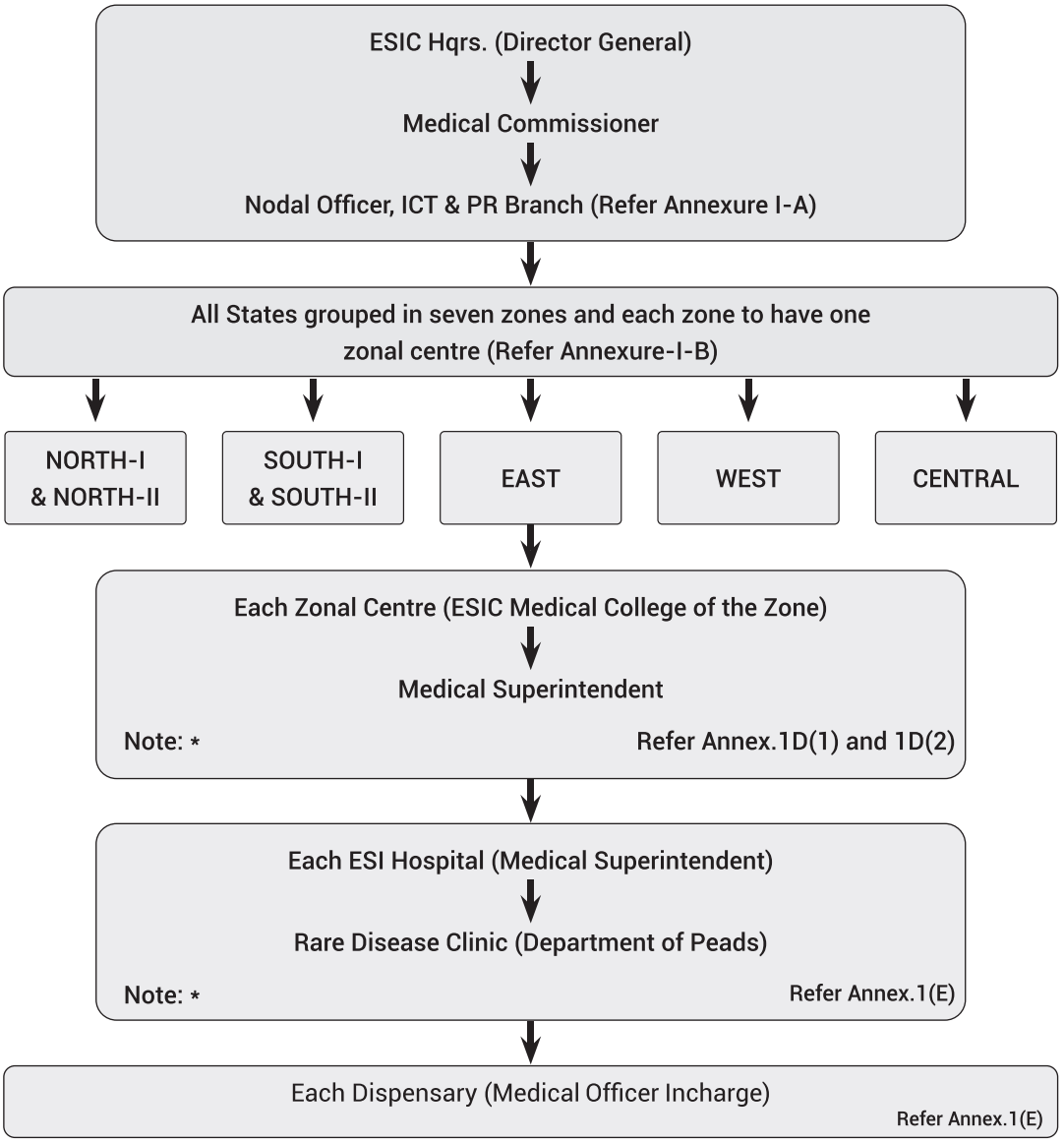
B Prepared a Teaching & Training Module for health care workers and IEC material for general public, which will be circulated shortly after a pilot run.

C A Dashboard is being created at ESIC Hqrs. for collection and compilation of data of rare diseases. Till the time it is made functional the field locations are required to collect and compile the data and forward to ESIC Hqrs. as per the above guidelines.

As field of rare disease is in nascent stage and the national policy is in the process of finalization the above structures & SOP's etc. may be revised as per feedback & requirement.

Annex.I

Organizational Structure for Rare Diseases in ESI



*In case of ESIS Hospitals the SMO of the State to coordinate with DIMS & MS ESIS Hospitals to facilitate management of cases of rare diseases.

Annex. IA

Role/Function of ESIC Hqrs. in R/O Rare Diseases

ESIC Hqrs. to nominate the Nodal Officer for Rare Disease

Nodal Officer

- Overall supervision and coordination with field locations.
- To coordinate between ESIC and MoHFW.
- Data collection and compilation.
- To create own Registry of ESIC for Rare Diseases with the objective of having a data base of various rare diseases from all field locations which will be forwarded to National Registry as and when created by ICMR as perNational Policy to avail the benefits.
- Updating the Rare Disease training modules.
- To resolve queries/grievances of ESI beneficiaries regarding rare diseases.

ICT Branch

- To make enabling provisions in Health Information System (HIS) forreporting and data entry of rare diseases.
- Provision for creating a pop-up facility for rare disease in the Dhanwantri Module.
- To create a rare disease dash board for ESIC to facilitate collection and forwarding rare disease data from field locations to ESIC registry.
- Creation of platform for patients to share their experiences and grievances.
- To create format for reporting of rare diseases on monthly basis from fieldlocations.

PR Department

- To prepare CD and posters on rare diseases from the material compiled by committee.
- To ensure availability of posters and booklets material.

Annex. IB

Zone wise distribution of States

Zone	State	Zonal Centre
North-1	Punjab, Chandigarh, Haryana & Uttar Pradesh	ESIC Medical College Faridabad
North-2	J&K, Himachal Pradesh Delhi & Uttrakhand	PGIMSR Basaidarapur Delhi
North-3	Rajasthan	ESIC Medical College Alwar, Rajasthan
South-1	Tamil Nadu, Puducherry & Kerala	ESIC Medical College & PGIMSR K.K. Nagar, Chennai
South-2	Karnataka & Andhra Pradesh	ESIC Medical College & PGIMSR Rajaji Nagar, Bangalore
East-1	West Bengal, Jharkhand, Assam-NER States & Odisha	ESI-PGIMSR, Medical College Joka, Kolkata
East-2	Bihar	ESIC Medical College Bihta, Patna, Bihar
West	Maharashtra & Goa	ESI PGIMSR Andheri, Mumbai
Central	Telangana, Chhatishgarh, Madhya Pradesh & Gujrat	ESIC Medical College Sanathnagar, Hyderabad

Annex. IC (1)

Zonal Centre (ESIC Medical College of the Zone)

ROLE/FUNCTIONS

DEAN (Overall supervision)

PSM Department

1. To organize and conduct trainingof trainers (Specialists, Medical Officers/ ANS/ DNS etc.) in the Zone by PSM Deptt. of Medical College in coordination withPaeds., obst, medicine and orthoDeptt.
2. To conduct, coordinate and supervise public awareness programmes through MS, State Centers & ESI Hospitals in the Zone.
3. To modify training module and awareness programme as per the need and requirement of the zone with approval from ESIC Hqrs.
4. Teaching institutes which do nothave the PSM department, the Peads. Deptt to take over the above function.

Nodal Officer*

1. Data collection of Rare Diseases from Rare Disease Clinics (MS, ESICH) through State Centres in the zone, its compilation, updation on the dashboard and reporting to Nodal Officer, ESIC Hqrs. For ESIS locations data be collected from SMO of the State.
2. To ensure collection, delivery and reporting of different dry blood samples rare disease clinics in their zone, once lab facilities are established.
3. To coordinate with State Govt., State Centre, rare disease clinics, Regional Office, Centre of Excellence, premier institutes and empanelled hospitals for facilitating the management of rare disease cases in the zone.
4. To manage the grievance redressal mechanism in coordination with State Centre.

Paeds. Department

1 To run the Rare Disease Clinic
2. To provide services for identification of high risk/ suspect cases, diagnosis, treatment and rehabilitation of confirmed cases, both to in house and referred from ESI dispensaries and linked states.
3. To provide/refer patients for optimum screening & diagnostic modalities to premier institutes / Centre of excellence for rare diseases (proposed)/ empanelled hospitals for confirmation of the disease (as per feasibility).
4. To send DBS samples to its Lab/Centre of excellence/ empanelled Centres.
5. To help the ESI hospitals in the zone to establish Rare Disease Clinics.
6. Monitoring of treatment & follow up of confirmed cases.
7. To ensure entry/entre the confirmed case in Dhanwantri Module.
8. To maintain record of expenditure made on diagnosis/ treatment of suspect/confirmed case and forward the same to Nodal Officer of the Zonal Centre for further transmission of data as per Annexure-5.

*Dean to depute supporting staff as per requirement.

Annex. IC (2)

Role/Function of Zonal Centre

Zonal Centre : Each Medical College in the zone to have a zonal centre

- Zonal Centre to function under overall supervision of Dean.
- Each Zonal Centre to cater to states linked to their Zone with respect to Rare Diseases.
- Zonal Centre also to function as State Centre, in case the hospital associated with College is the largest/Model ESIC hospital of the State.
- Dean to nominate a Nodal Officer: Faculty/ Specialist/ Medical Officer (additional responsibility).

Role and Functions of Nodal Officer

- Data collection of Rare Diseases from Rare Disease Clinics (MS, ESICH) through State Centres in the zone, its compilation, up-dation on the dashboard and reporting to Nodal Officer, ESIC Hqrs. For ESISlocations data be collected from SMO of the State.
- To ensure collection, delivery and reporting of different dry blood samples rare disease clinics in their zone, once lab facilities are established.
- To coordinate with State Govt., State Centre, rare disease clinics, Regional Office, Centre of Excellence, premier and empanelled hospitals for facilitating the management of rare disease cases in the zone.
- To manage the grievance redressal mechanism in coordination with State Centre.

Role and Functions of Paediatrics Department (Rare Disease Clinic)

To run the Rare Disease Clinic.

- To provide services for identification of high risk/suspect cases, diagnosis, treatment and rehabilitation of confirmed cases, both to in house and referred from ESI dispensaries and linked states.
- To provide/refer patients for optimum screening & diagnostic modalities to premier institutes / Centre ofexcellence for rare diseases (proposed)/ empanelled hospitals for confirmation of the disease.
- To send DBS samples to its Lab/Centre of excellence/ empanelled Centres.
- To help the ESI hospitals in the zone to establish Rare Disease Clinics.
- Monitoring of treatment & follow up of confirmed cases.
- To ensure entry/entre the confirmed case in Dhanwantri Module.

- To maintain record of expenditure made on diagnosis/ treatment of suspect/ confirmed case and forward the same to Nodal Officer of the Zonal Centre for further transmission of data as per Annexure-5.

Role and Functions of PSM Department (Training)

- To organize and conduct training of trainers (Specialists, Medical Officers/ ANS/ DNS etc.) in the Zone by in coordination with Paeds., Obst., Medicine and Ortho Dept. and to conduct, coordinate and supervise public awareness programmes through MS, State Centers & ESI Hospitals in the Zone.
- To modify training module and awareness programme as per the need and requirement of the zone with approval from ESIC Hqrs.
- Teaching institutes which do not have the PSM department, the Paeds. Dept. to take over the above function.

Annex. ID (1)

State Centre
(Largest ESIC Hospital/Model Hospital of the State)

ROLE/FUNCTIONS

Medical Superintendent

Nodal Officer*

- To collect data from Rare Disease Clinics (MS, ESICH) of the State/ compilation/ updation of data on dashboard and regular reporting to Nodal Officer, Zonal Centre.
- To ensure collection, delivery and reporting of different dry blood samples collected from rare disease clinics of their State in coordination with Nodal Officer of the Zonal Centre.
- To coordinate with Zonal Centre, Regional Offices, Rare Disease Clinics of hospitals, State Govt., Centre of Excellence, premier institutes, empanelled hospitals for facilitating the management of rare disease cases of the State, if required.
- To organize and conduct regular training of healthcare workers of the State (Hospitals & Dispensaries) and to conduct public awareness programmes through MS, ESI Hospitals of the state in coordination with Paeds., Obst., Medicine and Ortho Dept.
- To manage the grievance redressal mechanism in coordination with ESI Hospitals, for rare diseases.

Peads. Department

- To run the Rare Disease Clinic.
- To provide services for identification of high risk/suspect cases, diagnosis, treatment and rehabilitation of confirmed cases, both to in house and referred from ESI Hospital of the state.
- To provide/refer patients for optimum screening & diagnostic modalities to ESIC Medical College/premier institutes / Centre of excellence/empanelled hospitals for confirmation of the disease (as per feasibility).
- To send DBS samples to Lab at its Zonal Centre/ Centre of excellence for rare diseases (proposed)/ empanelled hospitals.
- To help the ESI hospitals in the State to establish Rare Disease Clinics.
- Monitoring of treatment & follow up of confirmed cases.
- To ensure that the confirmed case is entered in Dhanwantri Module.
- To maintain record of expenditure made on diagnosis/ treatment of suspect/ confirmed cases and forward to Nodal Officer of State Centre.

* MS to depute supporting staff as per requirement.

In case there is no ESIC hospital in the State, the RD/ SMO of the State will coordinate with Zonal Centre, DIMS, DMD/ DMN & MS ESIS Hospital of the State with respect to rare diseases.

Annex. 1D(2)

Role/Function of State Centre

Each state to have a **State Centre** in ESIC hospital of the largest/Model hospital of the State under supervision of MS, ESIC Hospital. State Centre to coordinate with ESI hospitals in the State & Zonal Centre of their Zone.

- MS to nominate a Nodal Officer: Specialist/ Medical Officer (additional responsibility) and to depute supporting staff as per requirement.
- In case, there is no ESIC Hospital in the State, then the State Medical Officer (SMO) to conduct trainingprogrammes for State ESI Hospitals and Dispensaries in coordination with the Dean of the Zonal Centre.

Role and Functions of Nodal Officer:

- To collect data from Rare Disease Clinics (MS, ESICH) of the State/ compilation/ up-dation of data on dashboard and regular reporting to Nodal Officer, Zonal Centre.
- To ensure collection, delivery and reporting of different dry blood samples collected from rare disease clinics of their State in coordination with Zonal Centre.
- To coordinate with Zonal Centre, Rare Disease Clinics of hospitals, State Govt., Centre of Excellence, premier institutes, empanelled hospitals for facilitating the management of rare disease cases of the State, if required.
- To organize and conduct regular training of healthcare workers of the State (Hospitals & Dispensaries) and to conduct public awareness programmes through MS, ESIC Hospitals.
- To manage the grievance redressal mechanism in coordination with ESI Hospitals, for rare diseases.
- To collect, compile, maintain the data pertaining to their institute and send the same to respective Nodal Officer of the State Centre.

A. Role and Functions of Paediatrics Department : Rare Disease Clinic (State Centre)

- To run the Rare Disease Clinic.
- To provide services for identification of high risk/ suspect cases, diagnosis, treatment and rehabilitation of confirmed cases, both to in house and referred from ESI Hospital of the state.

- To provide/refer patients for optimum screening & diagnostic modalities to ESIC Medical College/premier institutes / Centre of excellence/empanelled hospitals for confirmation of the disease (asper feasibility).
- To send DBS samples to Lab at its Zonal Centre/ Centre of excellence for rare diseases (proposed)/ empanelled hospitals.
- To help the ESI hospitals in the State to establish Rare Disease Clinics.
- Monitoring of treatment & follow up of confirmed cases.
- To ensure that the confirmed case is entered in Dhanwantri Module.
- To maintain record of expenditure made on diagnosis/ treatment of suspect/confirmed cases and forward to Nodal Officer of State Centre.

Annex. IE

Rare Disease Clinic

(All ESI Hospitals)

- Each ESI hospital to have a rare disease clinic, which will function under the overall supervision of MS and located in the Paediatrics department.
- MS to designate a Rare Disease Clinic Incharge .
- MS to update cases in the rare disease dashboard every month forward the data regarding rare disease to Nodal Officer (State Centre) .

Role and Functions of Rare Disease Clinic (Paediatrics Department)

- To provide services for identification of high risk/suspect cases, diagnosis, treatment and rehabilitation of confirmed cases, both to in house and referred from ESI dispensaries. • To provide/refer patients for optimum screening & diagnostic modalities to ESIC Medical College/premier institutes / Centre of excellence for rare diseases (proposed)/ empanelled hospitals for confirmation of the disease (as per feasibility). • To send DBS samples to Lab at its Zonal Centre/ Centre of excellence for rare diseases (proposed)/ empanelled hospitals for early & timely diagnosis (once relevant lab facilities are established at Zonal Medical College). • Monitoring of treatment & follow up of confirmed cases.
- To ensure entry/entre the confirmed case in Dhanwantri Module.
- To maintain record on expenditure made for diagnosis/ treatment of suspect/confirmed cases and to submit to their MS for onward transmission.
- To provide training of healthcare providers on rare diseases of ESI hospital and attached dispensaries in coordination with other departments (Obst., medicine and ortho Dept.).
- To generate awareness among ESI Beneficiaries.

Annex. IF

Role/function of Director (Medical)Delhi, Noida and State Directorates (Medical Services)

1. To ensure training of medical and para medical staff of ESI Dispensaries in coordination with MS ESI Hospitals (State Centre).
2. To direct Medical Officer Incharges for identification of high risk/suspected cases of rare diseases, their referrals to attached ESI Hospital for screening, early diagnosis and treatment.
3. To ensure of registration and entry of suspected/confirmed/follow-up cases of rare disease in Dhanwantry Module through ESI dispensaries (medical officer incharge).
4. To coordinate with attached ESI Hospital in respect of forwarding the rare disease data as per proforma provided.
5. To ensure training of medical and para medical staff through their Directorate in their area in coordination with State Centre and Zonal Centre and SMO.

Role/Function of Dispensaries

Medical Officer Incharge

1. At Dispensary Level Medical Officer Incharge to work under overall supervision of Director Medical of the State/Incharge Medical Services (DIMS) of the State, in respect to rare diseases in coordination with State Centre.
2. Referral of suspect/confirmed cases of rare diseases to rare diseases clinic of their attached ESI hospital.
3. Collection of rare disease data and forwarding to rare disease clinic/Nodal officer of attached ESI Hospital.

Annex. II

How to manage a Suspected case of Rare Disease

ESI Dispensary
(if suspected, including high risk i.e. with family history)

↓ Refer to

Attached ESI Hospital

↓

Refer to Concerned Deptt. for consultation and screening

↓

(Medicine, OBG, Paediatrics, Orthopaedics etc.)
[screening as per Annexure-6]

↓

If a rare disease is suspected in the speciality OPD

↓ Refer to

Rare Disease Clinic (under the department of Paediatrics)
for investigations to confirm the diagnosis

↓

Not diagnosed as rare disease, continue treatment at the concerned speciality

↓

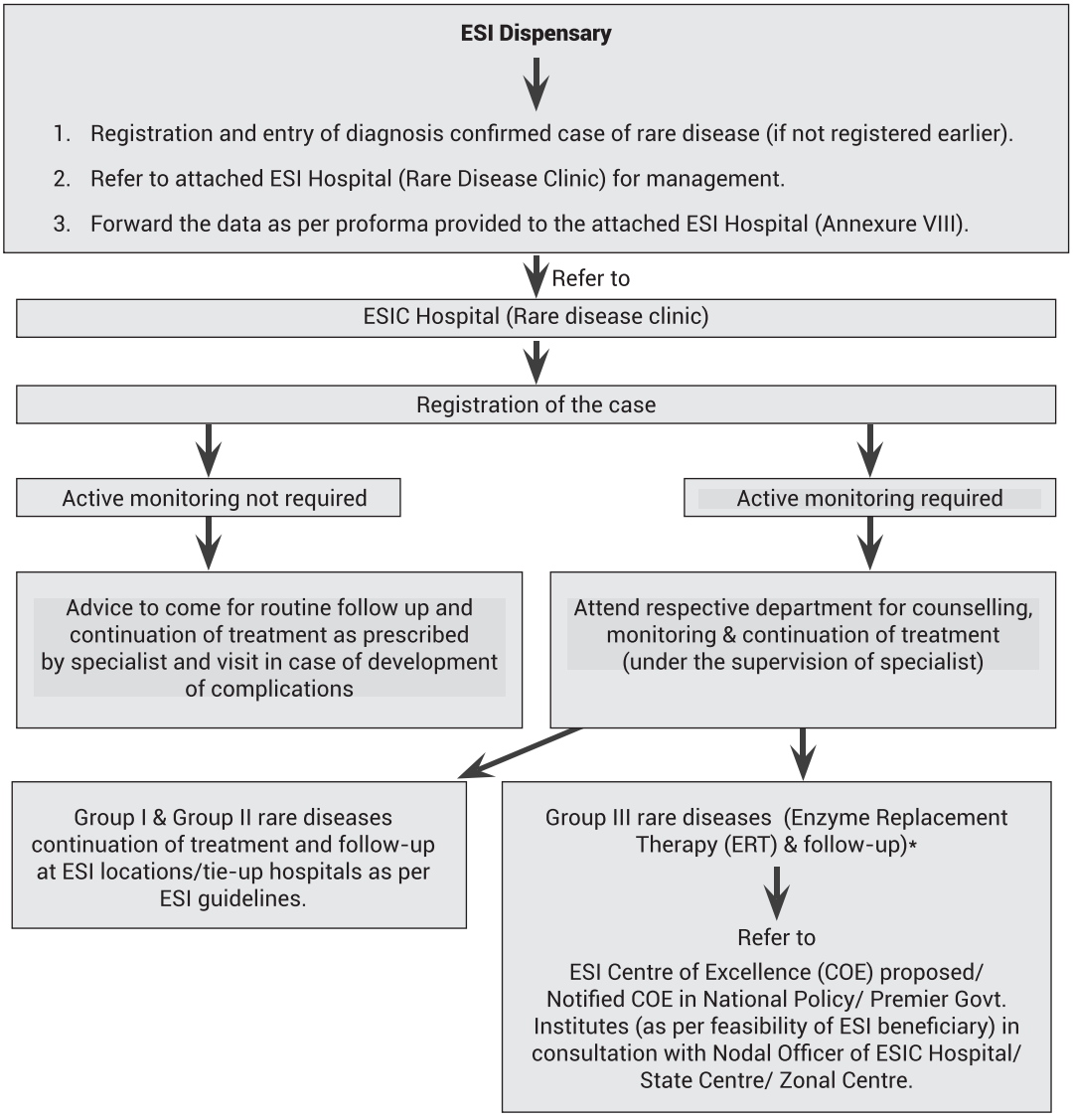
Confirmed Diagnosis of Rare Disease after Optimal Screening/ Diagnostic Test

↓

- To attend rare disease clinic for notification and further management:
  - a Counselling and treatment advice as per screening test.
  - b For Group I and II rare diseases for initiation of in-house treatment at the Zonal/ State Centre (if available) or at empanelled hospital.
  - c For Group III rare diseases- referral to designated Centre of Excellence for Rare Diseases (ESIC/ MOHFW)/ premier institutes in consultation with Nodal Officer of ESIC Hospital/State Centre/ Zonal Centre.

Annex. III

How to manage with a Confirmed case of Rare Disease



*Proforma/Diary for Enzyme Replacement Therapy (ERT) progress report to be maintained in the concerned ESI Hospital (Annexure VII A,B,C,D).

Annex. IV

Organisational Structure for training of Health Care Workers and awareness of public

Zonal Centre

↓

ESIC Medical College of the Zone (DEAN)

↓

PSM Department

- [In coordination with Paeds., OBG, Medicine Dept. etc. to organize and conduct regular training of trainers (Specialists, Medical Officers, Nursing Staff, ANMs etc.) of their Zone]. (Awareness also be extended to final year students)
- Attached ESI Hospital to organise public awareness programmes on regular basis under their supervision.

↓

State Centre

↓

Largest/Model ESIC Hospital (Medical Superintendent)

↓

- The Nodal officer (Incharge of training programme)
- [In coordination with Paeds., OBG, Medicine Dept. etc. to organize and conduct regular training of trainers (Specialists, Medical Officers, Nursing Staff, ANMs etc.) of their State] refer training module available in the link.
- ESI Hospital to also organise public awareness programmes on regular basis under their supervision(Refer CD/ Posters available in the link).

↓

ESIC Hospital Medical Superintendent

↓

1. The designated officer [in coordination with Paeds. Deptt. of ESI Hospital & State Centre to organize and conduct training of (Specialists, Medical Officers/Nursing Staff, ANMs etc.) of their hospital and attached dispensaries].

   1.ESI Hospital to also organise public awareness programmes on regular basis under their supervision.

   Note*

↓

Dispensary Level Medical Officer Incharge, To ensure training of all HCW at dispensary/ DCBO etc. in coordination with ESI Hospital/State Centre/ D(M)D/DIMS

Note: * in case there is no ESIC Hospital in the State then SMO to conduct training program for ESI Hospital and dispensaries in coordination with Zonal Centre.

Annex. V

(Organizational Structure for Data Collection & Compilation)

ESI Dispensary (Medical Officer Incharge)

A Entry of confirmed/follow-up case in Dhanwantri system.

B To maintain & forward the record to attached ESI hospital rare disease clinic through MS ESI Hospital (as per proforma Annexure VIII)

↓

State Centre (at largest/Model ESIC Hospital) Medical Superintendent

↓

Nodal Officer

1. Data collection from Rare Disease Clinics (MS, ESICH) of the State/ compilation/ updation of data on dashboard and regular reporting to Nodal Officer, Zonal Centre.
2. To ensure collection, delivery and reporting of different dry blood samples collected from rare disease clinics of their State in coordination with Zonal Centre.

↓

Rare Disease Clinic

1. To ensure that the confirmed case is entered in Dhanwantri Module.
2. To maintain record on expenditure made for diagnosis/ treatment of suspect/confirmed cases.
3. To update cases in the rare disease dashboard.
4. To collect, compile, maintain & send data to respective Nodal Officer.

Note*

↓

Zonal Centre (at ESI Medical College of the Zone) DEAN

Data collection of Rare Disease Clinics (MS, ESICH) through State Center of the Zone/ compilation/ updation of data on dashboard and regular reporting to Nodal Officer, ESIC Hqrs.

Rare Disease Clinic

1. To ensure that the confirmed case is entered in Dhanwantri Module.
2. To maintain record on expenditure made for diagnosis/ treatment of suspect/ confirmed cases.
3. To update cases in the rare disease dashboard.
4. To collect, compile, maintain & send data to respective Nodal Officer.

ESIC Hqrs. (Medical Commissioner)

↓

Designated Nodal Officer

↓

1. To ensure timely receipt of data from all Zonal Centres, compilation, confirmation of updation of data in rare disease dashboard and onward transmission to Central Registry at ICMR as per National Policy.
2. Data analysis and further needful action.

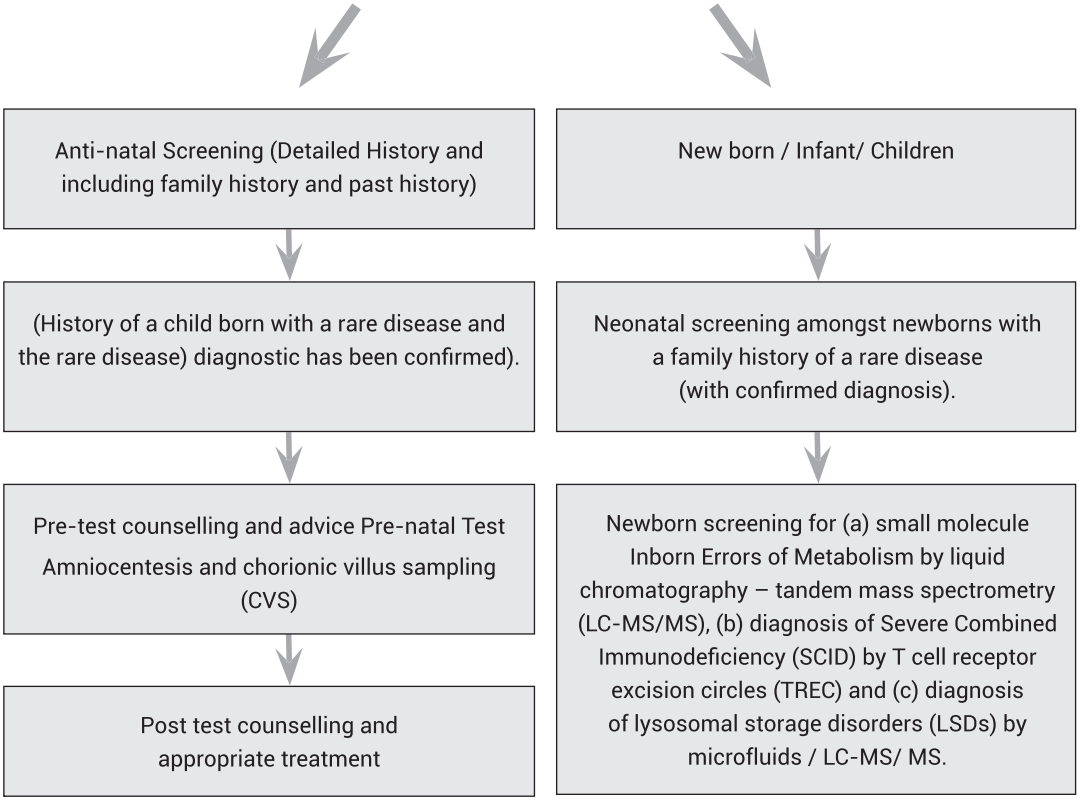
Note: *In case of non -existence of ESIC Hospital in the State, DIMS in coordination with SMO, to ensure data collection, compilation and onward transmission to Zonal Centre.

State and Zonal Centres in coordination with SMO of the state to ensure data collection, compilation , up-dation in dashboard and coordination for timely procurement and availability of treatment for ESIS hospitals & dispensaries.

The overall supervising authority of the locations to ensure mechanism for coordination of all departments for timely collection and compilation of information as per formats so that no such case of rare diseases are left out.

Annex. VI

Optimal Screening and Diagnosis of Rare Disease



As preventive measure, optimum screening and diagnosis, based on the directions suggested in the National Policy i.e. "those pregnant women in whom there is a history of a child born with a rare disease and that rare disease diagnosis has been confirmed, would be offered prenatal screening test(s) through amniocentesis and / or chorionic villi sampling. In case, the diagnosis could not be established during the prenatal period, it would be offered to the new born or the infant as the case may be through new born screening tests".

Antenatal card should include following points in history relevant to rare disease:

a) Chromosomal disorders, e.g. Down syndrome
b) Mental retardation or intellectual delay
c) Family/past obstetric history of congenital malformations, e.g. congenital heart defects, neural tube defect, and orofacial clefts.
d) Unexplained early neonatal death.
e) Known genetic disorder in family, e.g. thalassemia, sickle cell anemia, bleeding disorders.
f) Deafness, Recurrent miscarriages. Familial cancer syndromes (known or suspected).

Annex VII A1

Proforma/Diary for Enzyme Replacement Therapy (ERT) to be maintained in ESI Hospital (Gaucher's Disease)

Photo of Patient

Name of Patient ..... Name of IP..... Insurance No. ..... Date of Birth of patient ..... Sex ..... Diagnosis ..... Date of Diagnosis..... Date of start of ERT..... ERT advised by (Name of Hospital).....

Sl. No.	Date of ERT	Name of Drug	Dose	Clinical status as on date Improving/same/ deteriorating	Identityverified	Name & Signature of consultant

NOTE:

- Signature of consultant to be done on every visit.
- Signature of Medical Superintendent at the time of sanction of the drug after ensuring correctness of the details filled in andto be forwarded to ESIC Hqrs. at the time of referring the case to High Cost Treatment Committee.
- To be maintained at the referring ESI hospital (in concerned department & store) on a hard paper/ sheet/folder. The samemay be asked by ESIC Hqrs. Office at any time.

(Medical Superintendent)

Annex. VII A2

Clinical assessment for Gaucher's disease (At baseline, 6 months and thereafter yearly)

1	2	3	4	5	6	7	8	9	10	11	12
Date	Weight	Height	Liver	Spleen	Hemogram	Dexa	MRI Spine & Femoral Neck	Chito-triosidase	PET	HRCT	2D Echo

- Monitoring of Sl. No. 2, 3, 4 & 5 to be done every 6 months.
- Monitoring of Sl. No. 7, 8 & 9 to be done yearly.
- Monitoring of Sl. No. 10, 11 & 12 to be done as per indication.

(Name and Signature of Consultant)

Annex. VII C1

Proforma/Diary for Enzyme Replacement Therapy (ERT) to be maintained in ESI Hospital (Fabry Disease)

Photo of Patient

Name of Patient ..... Name of IP..... Insurance No. ..... Date of Birth of patient ..... Sex ..... Diagnosis ..... Date of Diagnosis..... Date of start of ERT..... ERT advised by (Name of Hospital).....

Sl. No.	Date of ERT	Name of Drug	Dose	Clinical status as on date Improving/same/ deteriorating	Identityverified	Name & Signature of consultant

NOTE:

- Signature of consultant to be done on every visit.
- Signature of Medical Superintendent at the time of sanction of the drug after ensuring correctness of the details filled in and to be forwarded to ESIC Hqrs. at the time of referring the case to High Cost Treatment Committee.
- To be maintained at the referring ESI hospital (in concerned department and store) on a hard paper/sheet/folder. The same may be asked by ESIC Hqrs. Office at any time.

(Medical Superintendent)

Annex. VII C2

Clinical assessment for Fabry Disease
(To be forwarded to Hqrs. every 6 months)

1	2	3	4	5	6	7	8	9	10	11
Date	Weight	Height	Symptoms (specify)	Haemogram	KFT/ S.Elec	Urine Protein	Hearing/Vision assessment	Cardiac status	CNS	others

- Monitoring of 2,3,4,9,10 to be done on every visit.
- Monitoring of 5,6,7 to be done on base line and monthly visit.
- Monitoring of 8 and 11 to be done on base line and as required.

(Name and Signature of Consultant)

Annex. VII B-1

Proforma/Diary for Enzyme Replacement Therapy (ERT) to be maintained in ESI Hospital Mucopolysacharidosis (MPS)

Photo of Patient

Name of Patient ..... Name of IP.....
Insurance No..... Date of Birth of patient..... Sex .....
Diagnosis..... Date of Diagnosis .....
Date of start of ERT.....ERT advised by (name of hospital).....

Sl. No.	Date of ERT	Name of Drug	Dose	Clinical status as on date Improving/same/ deteriorating	Identity verified	Name & Signature of consultant

NOTE:

- Signature of consultant to be done on every visit.
- Signature of Medical Superintendent at the time of sanction of the drug after ensuring correctness of the details filled in and to be forwarded to ESIC Hqrs. at the time of referring the case to High Cost Treatment Committee.
- To be maintained at the referring ESI hospital (in concerned department and store) on a hard paper/sheet/folder. The same may be asked by ESIC Hqrs. Office at any time.

(Medical Superintendent)

ANNEX. VII B-2

Clinical assessment for MPS
(At baseline, 6 months and thereafter yearly)

1	2	3	4	5	6	7	8	9	10
Date	Weight	Height	Liver	Spleen	Snoring	6 minute walk test	3 minute stair climb test	Cardiac Status	Ophthalmological Examination

	11	12	13	14	15	16
Date	PFT FEV1 FVC FEV1/FVC PEFR (Pred%) (Pred%) (Pred%) (Pred%)	Range of movements shoulder flexion, elbow extension, knee extension	Polysomnography	HRCT Chest	2D Echo	Others

- Monitoring of 2 to 6 to be done every 6 monthly.
- Monitoring of 7 to 12 to be done yearly.
- Monitoring of 13, 14 & 15 to be done as per indication.

(Name and Signature of Consultant)

Annex. VII D

Proforma/Diary for Enzyme Replacement Therapy (ERT) to be maintained in ESI Hospital (other than MPS/Gaucher's/Fabry's)

Photo of Patient

Name of Patient ..... Name of IP.....

Insurance No..... Date of Birth of patient..... Sex .....

Diagnosis..... Date of Diagnosis .....

Date of start of ERT ..... ERT advised by (name of hospital).....

Sl. No.	Date of ERT	Name of Drug	Dose	Clinical status as on date Improving/same/ deteriorating	Identity verified	Name & Signature of consultant

NOTE:

- Signature of consultant to be done on every visit.
- Signature of Medical Superintendent at the time of sanction of the drug after ensuring correctness of the details filled in and to be forwarded to ESIC Hqrs. at the time of referring the case to High Cost Treatment Committee.
- To be maintained at the referring ESI hospital (in concerned department & store) on a hard paper/sheet/folder. The same may be asked by ESIC Hqrs. Office at any time.

(Medical Superintendent)

Annex. VIII

Format for entry of details in the dashboard of rare diseases (to be updated in the first week of every month)											
1	2	3	4	5	6	7	8	9			
Name of the Location	ESIC location Code Number	Date and month of entry in dashboard	Name of the patient	Name of the IP	Insurance Number	Gender	Age & relationship with IP	Diagnosis			
								Group	Disease	Suspected	confirmed
								Group 1: Disorders amenable to one-time curative treatment:			
								a)	Disorders amenable to treatment with hematopoietic stem cell transplantation (HSCT)-		
								a.	Such Lysosomal Storage Disorders (LSDs)		

10	11	12	13	14	15	16	17
Date of start of treatment of confirmed case	Total Expenditure on treatment for confirmed case at the end of financial year.	No of suspected case of Group 1 in the Zone (to be filled by Nodal Officer Zonal Centre)	No of suspected case of Group 2 in the Zone (to be filled by Nodal Officer Zonal Centre)	No of suspected case of Group 3 in the Zone (to be filled by Nodal Officer Zonal Centre)	No of confirmed case of Group 1 in the Zone (to be filled by Nodal Officer Zonal Centre)	No of confirmed case of Group 2 in the Zone (to be filled by Nodal Officer Zonal Centre)	No of confirmed case of Group 3 in the Zone (to be filled by Nodal Officer Zonal Centre)

18	19	20	21	22	23
Expenditure incurred on diagnosis of suspected case of Group 1 (to be filled by Nodal officer Zonal Centre)	Expenditure incurred on diagnosis of suspected case of Group 2 (to be filled by Nodal officer Zonal	Expenditure incurred on diagnosis of suspected case of Group 3 (to be filled by Nodal officer Zonal Centre)	Expenditure incurred on treatment of confirmed case of Group 1 (to be filled by Nodal officer Zonal Centre)	Expenditure incurred on treatment of confirmed case of Group 2 (to be filled by Nodal officer Zonal Centre)	Expenditure incurred on treatment of confirmed case of Group 3 (to be filled by Nodal officer Zonal Centre)

Data be compiled and sent to ESIC Hqrs. (Nodal Officer Rare Diseases) as per the proposed format above, on monthly basis in hard copy by the MS ESIC Hospitals, Nodal Officer of the State and Zonal Centres, SMO (for ESIS Beneficiaries in coordination with State Directorates).

NOTE

NOTE

क.रा.बी.नि
E S I C

कर्मचारी राज्य बीमा निगम
Employees' State Insurance Corporation
श्रम एवं रोजगार मंत्रालय
भारत सरकार
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