

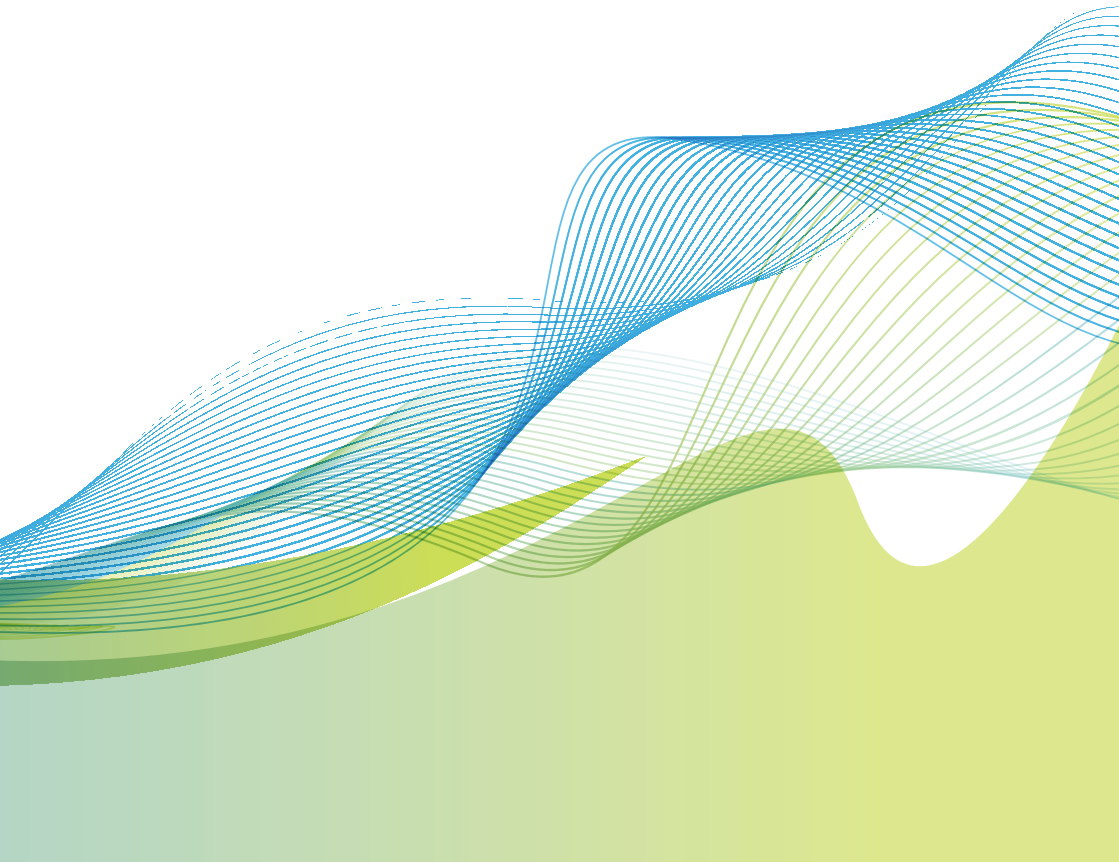


KEMENTERIAN KESIHATAN MALAYSIA

MALAYSIAN ORPHAN MEDICINES Guideline

PROGRAM PERKHIDMATAN FARMASI
KEMENTERIAN KESIHATAN MALAYSIA

MALAYSIAN ORPHAN **MEDICINES** Guideline



MALAYSIAN ORPHAN MEDICINES GUIDELINE

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ACKNOWLEDGEMENT

The Ministry of Health would like to express its sincere gratitude to all who have contributed to the success of the publication of this guideline.

To all the committee members from the public and private sector, the Ministry would like to congratulate and convey its appreciation for pouring in efforts and time. Your commitment in preparing this guideline will bring a change to the orphan medicine industry towards providing better health outcomes for the nation.

Good support and advocacy by the patient support group have benefited particularly the patients and their families. Their enthusiasm and engagements have facilitated the Ministry throughout the preparation of this guideline.

The Ministry also wishes to record its appreciation to those who have participated in any capacity to accomplish this guideline. The many contributions will always be remembered.

With this, the Ministry hopes to obtain continuous support from all in promoting and implementing this guideline for the benefit of the whole nation.

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- Malaysian Association of Pharmaceutical Suppliers (MAPS)
- Malaysian Rare Disorders Society (MRDS)
- Malaysian Lysosomal Disease Association (MLDA)
- Malaysian Metabolic Society (MMS)
- WeCareJourney

FOREWORD OF THE DIRECTOR-GENERAL OF HEALTH



A rare disease is a condition that affects small numbers of the population and is also known as an orphan disease in some countries. Globally, there are more than 8,000 types of rare diseases documented and notably, the definition of rare diseases varies from one country to another due to the local distinction or unknown prevalence of rare diseases. In Malaysia alone, there are over 500 recognised diseases currently registered on the Malaysian Rare Disease List.

Rare diseases are profoundly different from common chronic diseases and challenging to clinicians in diagnosing, treatment and providing care. Scarce resources in medical expertise, fragmented disease knowledge, the lack of advanced equipment, and dedicated infrastructures are the critical factors contributing to the complexity of managing the rare disease. Beyond these hurdles, sustainable financing from the government is vital to ensure the care and access to diagnosis and treatment are available for patients who required it the most.

The Ministry of Health acknowledges the needs of patients with a rare disease in Malaysia. Rising to these demands, the National Framework for Rare Disease developed to deliver integrated, comprehensive, and holistic management for rare disease patients. This framework unified all the stakeholders and paved the way for consolidated policies and strategies in ensuring that affected people can access the services they need, when and where they need them, and possibly reduce the financial burden. Despite the challenges in the implementation of a holistic national programme for rare diseases, the Ministry believes active participation and partnership from all stakeholders will realise the vision of the 'Nation Working Together for Better Health'.

The official publication of documented guidelines related to orphan medicines published under this national framework is timely. It is a concerted effort between various divisions in the Ministry of Health, pharmaceutical industries, academicians, and non-governmental organisations that should be acknowledged as a stepping stone towards further collaborations to improve the nation's health, notably in strengthening the rare disease treatment implementation network.

I would like to express my appreciation and congratulation to the Pharmaceutical Services Programme for their diligence and effort in securing the initiative under the Malaysian National Rare Disease Framework. My sincere gratitude extends to all stakeholders and their contributions and support to materialised this guideline and further enhanced the policies. Let us continue to work together and leave no one behind!

Thank you.



TAN SRI DATO' SERI DR. NOOR HISHAM BIN ABDULLAH
Director-General of Health Malaysia



FOREWORD OF THE SENIOR DIRECTOR OF PHARMACEUTICAL SERVICES



Pharmaceutical Services Programme is committed to ensure accessibility of medicines in Malaysia including medicines to treat rare diseases. Under the Malaysian National Medicines Policy (MNMP) or *Dasar Ubat Nasional* (DUNas), one of the key initiatives is to enhance the accessibility of life-saving products and orphan medicines without compromising safety, quality and efficacy of the medicines. Being an integral part of a health system, pharmaceutical services must be able to stay ahead and keep up with the demand to ensure that ‘no one is left

behind’ as pledged in 2030 Sustainable Development Goals (SGDs).

This is the first officially documented guideline related to orphan medicines that is published by the Programme. The publication of this guideline shows the commitment of the Pharmaceutical Services Programme in enhancing the accessibility of orphan medicines which emphasise on the facilitation of access through regulatory pathway. Procedures related to designation, regulation, post marketing surveillance and availability of orphan medicines are addressed in this guideline in order to ensure non-commercially viable products can be made available without compromising quality and safety of the medicines. This guideline also serves as a reference for the healthcare providers in handling the use of orphan medicines in the healthcare facilities.

Therefore, I would like to congratulate all the individuals involved in completing this guideline. On behalf of the Pharmaceutical Services Programme, I would also like to express my heartfelt gratitude to all the committee members, various agencies in the Ministry of Health, pharmaceutical industries, patient support group and other stakeholders for the continuous effort and valuable contributions towards publication of this guideline.

I believe, with the collaborated effort from all and continuously working hand in hand to fill in the gaps in rare disease treatment, may successfully overcome the challenges and issues in the future. Let us work together to achieve equal health for the nation.

Thank you.

DATIN DR. FARIDAH ARYANI BINTI MD. YUSOF
Senior Director of Pharmaceutical Services
Ministry of Health Malaysia

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GLOSSARY OF TERMS, ABBREVIATIONS AND ACRONYMS

ADR	Adverse Drug Reaction
Applicant	Registered Company
ATC	Anatomical Therapeutic Chemical
BPF	<i>Bahagian Perkhidmatan Farmasi</i> (new: <i>Program Perkhidmatan Farmasi</i>)
COA	Certificate of Analysis
CPP	Certificate of Pharmaceutical Product
DCA	Drug Control Authority
DEC	Drug Evaluation Committee
DRGD	Drug Registration Guidance Document
DUNas	<i>Dasar Ubat Nasional</i>
EU	European Union
FUKKM	<i>Formulari Ubat Kementerian Kesihatan Malaysia</i>
GMP	Good Manufacturing Practice
KP	<i>Kad Pengenalan</i>
MAL	Malaysia
MNMP	Malaysian National Medicines Policy
MOH	Ministry of Health
NPRA	<i>Bahagian Regulatori Farmasi Negara</i> (or National Pharmaceutical Regulatory Agency)
PBKD	<i>Pihak Berkuasa Kawalan Dadah</i>
PRH	Product Registration Holder
PSP	Pharmaceutical Services Programme
RM	<i>Ringgit Malaysia</i>
SDGs	Sustainable Development Goals

1.0 RARE DISEASES AND ORPHAN MEDICINE IN MALAYSIA

Rare diseases, as described in the European Union (EU), are diseases which affect a small number of people compared to the general population and specific issues are raised in relation to their rarity. Over six to seven thousand rare diseases have been discovered and these diseases are serious, often chronic and progressive. There is no cure for most rare diseases, but appropriate treatment and medical care can improve the quality of life of those affected and extend their life expectancy. Scientific and medical knowledge which in the field is rare, rendered the patients undiagnosed, misdiagnosed or with delayed treatment.

The Ministry of Health (MOH) Malaysia recognises the needs of the patients with rare diseases in Malaysia. Under the Malaysian National Medicines Policy (MNMP) one of the focus areas to improve on is to enhance the accessibility of life-saving products and orphan medicine without compromising safety, quality and efficacy of the medicines.

The purpose of this guideline is to facilitate access to medicines required for the management of rare diseases in the context of the Ministry, based on a consensus list of rare diseases compiled through consultation with rare disease experts.

1.1 Objectives

- To improve access to safe, quality and efficacious orphan medicine for the treatment of rare diseases.
- As a guide for healthcare professionals who manage rare diseases and industries, especially with regard to ensure continuity and effectiveness of the treatment.

1.2 Definitions

- **Orphan Medicine**

A medicinal product that is primarily intended to treat, prevent or diagnose a rare disease.

- **Rare Disease**

A life-threatening and / or chronically debilitating rare condition as listed in the Malaysian Rare Disease List (Appendix A)

1.3 Categorisation of Orphan Medicine

Orphan medicine shall be categorised according to its use.

1.3.1 Emergency Treatment

- Immediate intervention for life-saving / avoidance of permanent disability.
- For accessibility, the medicines shall be available as ready inventory (stock keeping) and shall be managed as emergency item.

1.3.2 Lifetime treatment

- Lifetime treatment is defined as long term treatment and maintenance therapy for rare disease.
- The medicines may be in either pharmaceutical dosage forms or non-pharmaceutical dosage forms (e.g. powder and liquid in the form of raw material).
- The orphan medicine shall be made available accordingly by the facilities that managed the patients.

2.0 DESIGNATION AND REGISTRATION OF ORPHAN MEDICINE

2.1 Designation of Orphan Medicine

The designation of orphan medicine is under the purview of *Bahagian Regulatori Farmasi Negara* (NPRA) through input from the Drug Evaluation Committee (DEC).

2.1.1 Eligibility Criteria for the Designation of Orphan Medicine

The designation of orphan medicine is subject to the following criteria:

- a) "A medicine, vaccine or in vivo diagnostic agent that is primarily intended to treat, prevent or diagnose a rare disease"¹; and
- b) No satisfactory method of diagnosis, prevention or treatment of the condition concerned can be authorised; or, if such a method exists, the medicinal product must be of significant benefit² to those affected by the condition.

2.1.2 Application for the Designation of Orphan Medicine

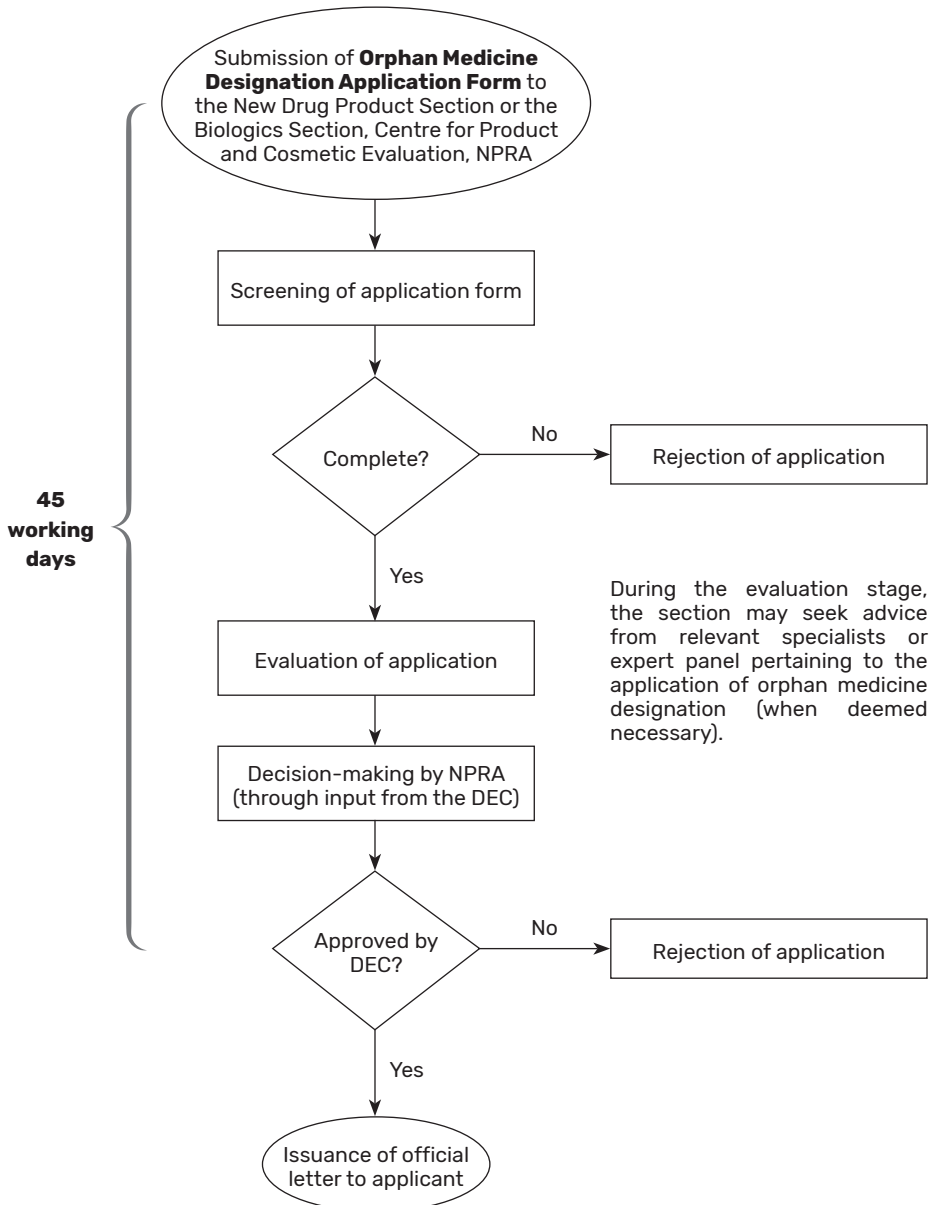
- a) The applicant may submit an application for such designation to the NPRA using the **Orphan Medicine Designation Application Form** as in **APPENDIX B** which can also be downloaded from the NPRA official website at <https://www.npra.gov.my/>.
- b) The application can be submitted before a product is registered as a New Chemical Entity or a Biologic product.
- c) The information required for the application of orphan medicine designation may include but are not limited to the following:
 - i) Product Information
 - Product name
 - Active ingredient
 - Strength
 - ATC Code
 - Pharmaceutical form
 - Route of administration
 - Manufacturer name and address
 - Worldwide regulatory status
 - Worldwide orphan medicine designation status

¹Rare disease refers to the diseases listed in latest Malaysian Rare Disease List.

²Significant benefit means that a medicine produces a clinically relevant advantage or makes a major contribution to patients' care, compared with existing methods to treat the condition. Thus, orphan designation is given to a product that will improve patients' current treatment, having considered what else is available¹⁰.

- ii) Proposed Rare Disease and Condition
 - Proposed Indication related to the rare disease
 - Brief Description of the rare disease
 - Current available method in treating/preventing/ diagnosing the rare disease
 - Justification for this product to be designated as orphan medicine
 - Brief description of the product (details on active ingredient(s), medicines type/class, structure, physical-chemical properties)
 - Mechanism of action explaining how the product works in relevant disease/condition
- iii) Scientific rationale for the orphan medicine use (the scientific rationale should support a medical plausible basis for the orphan medicine to be effective in treating disease/condition)
 - Please briefly describe the scientific evidence to support safety and efficacy of this product to treat/prevent/ diagnose the proposed indication related to the rare disease
 - Tabulated pre-clinical data and clinical data
 - A brief safety update report
- d) A medicinal product that has already been granted an orphan medicine designation in other countries is not automatically designated as an orphan medicine in Malaysia. It is still subject to the decision from NPRA (through input from the DEC).
- e) The same medicinal product may also have multiple orphan medicine designations for different rare diseases.
- f) NPRA may seek advice/opinion from relevant experts or representatives from rare disease society/patient groups or other key opinion leaders pertaining to the application of orphan medicine designation when deemed necessary.

2.1.3 Application Workflow and Procedure for Designation of Orphan Medicine



2.1.4 Cancellation of Orphan Medicine Designation

- The NPRA (through input from the DEC) may, at any time and by notice, cancel any orphan medicine designation of an unregistered/registered medicinal product that no longer meets the criteria for such designation.
- However, the registration status of that medicinal product shall remain valid.

2.2 Registration of Orphan Medicine

Note: This part shall be read in conjunction with the current DRGD

2.2.1 Procedure, Fee and Timeline

Procedure, fee and timeline for registration of orphan medicine shall follow those stated in the DRGD.

2.2.2 Registration Requirements and Conditions

Certain flexibilities are permitted for the registration of new medicinal products (i.e. new chemical entities and biologics) as orphan medicine as described in the DRGD.

2.2.3 Listing of DCA Registered Orphan Medicine

A list of orphan medicine registered with the DCA shall be published on the NPRA official website. The list will be updated regularly as and when updates have been made to it.

2.2.4 Re-Registration

The re-registration procedure stated in the DRGD shall apply to the re-registration of an orphan medicine.

2.2.5 Cancellation of Orphan Medicine Registration

The DCA may, cancel any registered orphan medicine that no longer meets the criteria for registration.

The cancellation procedure/details stated in the DRGD shall apply to the cancellation of a registered orphan medicine.

3.0 ACCESSIBILITY OF MEDICINE TO TREAT RARE DISEASE

Accessibility of medicine plays a key role in determining whether patient will receive adequate and effective treatment. The primary goal is to create an appropriate mechanism to facilitate the availability of medicine to treat rare disease.

3.1 Availability of Medicine

When the medicine required to treat rare disease has to be imported from outside the country, prescriber shall obtain permission from the MOH prior to getting supplies to assist patients in receiving alternative treatment or early access to treatment at both the public and private healthcare institutions.

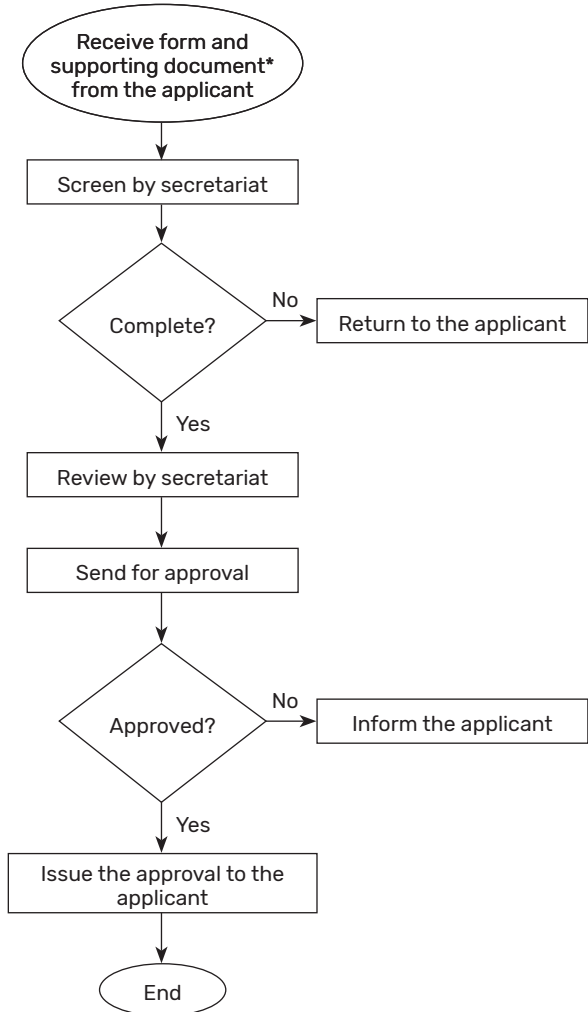
3.1.1 Application

Application for importation of medicine to treat life-threatening rare disease shall be submitted by the healthcare provider or any appointed company that has been awarded by the government contract to the Pharmaceutical Services Programme, MOH. The relevant forms and supporting documents shall be submitted:

- i) Applicants from the MOH institutions are required to use the **'Medicines Approval Form' / 'Permohonan Memperoleh Dan Menggunakan Ubat Yang Memerlukan Kelulusan Khas (For MOH Facilities)' Form** as in **Appendix C**.
- ii) Applicants from other public institutions or private institutions are required to use the **'Application to Import/Manufacture Unregistered Products for the Treatment of Life-Threatening Illnesses (For Private/ Non-MOH Institutions)' Form** as in **Appendix D**.

All healthcare facilities shall adhere to the above-mentioned procedures in supplying medicines to treat rare disease to patients. All forms and guidelines are available through the official website of the Pharmaceutical Services Programme at www.pharmacy.gov.my.

3.1.2 Workflow and Procedure of Application for Medicines of Special Approval / Application to Import Product for the Treatment of Life-Threatening Illnesses



***Supporting documents (applicable for company):**

- 1. GMP Certificate**
- 2. Finished Product Certificate of Analysis (CoA)**
- 3. Certificate of Pharmaceutical Product (CPP)**
- 4. Product label artwork**
- 5. Product Package Insert**
- 6. Clinical evidences (if any)**

* A minimum of three (3) of these documents are required as supporting documents

4.0 POST-MARKETING ACTIVITIES

All registered orphan medicine used in Malaysia shall be subjected to post-marketing activities. As such, the PRH shall be the responsible entity to implement the requirements.

The PRH shall appoint a responsible person in handling post-marketing issues in Malaysia. The details of the current responsible person, such as name, postal address, e-mail address, telephone, and fax numbers shall be provided to the NPRA and are required to be promptly informed if there is any change.

4.1 Surveillance and Product Complaint

The requirement for registered product quality monitoring is described in the DRGD.

4.2 Pharmacovigilance

The PRH is responsible to ensure that an appropriate system of pharmacovigilance is in place. The PRH shall continuously monitor and determine whether benefits continue to outweigh risks, and to consider the necessity for steps to improve the benefit-risk balance through risk minimization activities. The PRH is responsible and liable for their products on the market and must take appropriate actions, when necessary.

For full details on the requirements related to pharmacovigilance, please refer to the *Malaysian Pharmacovigilance Guidelines*.

i) Management of adverse drug reaction (ADR)

The PRH should have in place written procedures describing the handling of all ADRs related to their products. The system and procedures in place must be adequate for receipt, handling, evaluation and reporting of ADRs to the NPRA within the stipulated timelines stated in *Malaysian Pharmacovigilance Guidelines*.

ii) Annual submission of safety reports

Submission of Periodic Safety Updates Report / Periodic Benefit Risk Evaluation Report every 6 months for the first 2 years and once a year for the following 3 years is required for new drug products and biologic products.

If the requirements cannot be fulfilled, the PRH shall provide an annual safety report, which includes:

- A summary (line listing and summary tabulation) report of all the ADR cases received during a period of twelve months. It is preferably submitted in the PRH ADR Summary Report format defined in the *Malaysian Pharmacovigilance Guidelines*.
- A review of ongoing clinical study.

- Any risk minimization activities or programmes requested by other regulatory authorities relevant to the registered orphan medicines.
- A description of the investigation plan for the coming year.

iii) Emerging safety issues

Events/observations related to a registered orphan medicine may occur, which may have major impact on the risk-benefit balance of the product and/or on patients or public health. They may require urgent attention of the DCA and could warrant prompt regulatory action and communication to patients and healthcare professionals. These important new evidences should be considered as emerging safety issues. The PRH shall:

- alert the NPRA of any emerging global safety issue(s)
- submit all relevant safety information such as post-registration study reports and risk management plan as instructed by the NPRA
- respond promptly to the NPRA on request for additional risk-benefit information of the products involved

Any emerging safety issue shall be notified to the NPRA within the stipulated timeline stated in the *Malaysian Pharmacovigilance Guidelines*.

iv) Safety communication

Please refer to the *Malaysian Pharmacovigilance Guidelines* for further detail.

5.0 RESPONSIBILITY OF HEALTHCARE FACILITIES

All healthcare facilities shall be responsible in ensuring accessibility and availability of orphan medicines used in their facilities. In order to ensure availability of orphan medicines and continuity of treatment, all healthcare facilities treating rare diseases are encouraged to ensure orphan medicines are readily available and in adequate quantity for patients' use, subject to resources availability.

Patient monitoring is required for all products for their safety and efficacy including products that have been approved through special approval by Senior Director of Pharmaceutical Services.

All healthcare professionals should report any suspected ADR related to treatment with orphan medicines. ADR reporting can be done via the official website of NPRA at www.npra.gov.my - click on '**Report ADR as a healthcare professional.**'

FOR MORE INFORMATION

Please contact the respective department for further enquiry:

- **DESIGNATION AND REGISTRATION OF ORPHAN MEDICINE:**

Industry Development and Communication Section (One Stop Centre)
Bahagian Regulatori Farmasi Negara (NPRA)

Tel : 03-7883 5400 ext. 4437/4436/4435

Fax : 03-7956 7075

E-mail : npra@npra.gov.my

- **APPLICATION TO IMPORT PRODUCT TO TREAT RARE DISEASE:**

Formulary Management Subdivision
Pharmacy Practice and Development Division
Ministry of Health Malaysia

Tel : 03-7841 3200

Fax : 03-7968 2222

- **MARKET SURVEILLANCE / PHARMACOVIGILANCE / ADVERSE DRUG REACTION (ADR) REPORTING:**

Centre for Compliance and Quality Control
Bahagian Regulatori Farmasi Negara (NPRA)

Tel : 03-7883 5441

E-mail : fv@npra.gov.my (ADR Report)

E-mail : qpr@npra.gov.my (Product Quality Reporting)

- **MALAYSIAN RARE DISEASE LIST:**

Obstetrics & Gynaecology, Paediatrics Services Unit
Medical Development Division
Ministry of Health Malaysia

Tel : 03-8883 1047

Fax : 03-8883 1427

APPENDIX A: MALAYSIAN RARE DISEASE LIST

(Preliminary list as of August 2020)

Definition:

Rare disease is defined as a life-threatening and/or chronically debilitating rare condition as listed in the Malaysian Rare Disease List.

The criteria for inclusion of a disease in the Malaysian Rare Disease List are: -

1. There are or have been confirmed patients in Malaysia
2. The disease affects fewer than 1 in 4,000 people in Malaysia*
3. The disease is a severe condition
4. Its inclusion is approved by the national advisory committee**

* *Based on expert opinions and on local epidemiologic data where available*

** *National Rare Disease Committee (Jawatankuasa Penyakit Jarang Jumpa Kebangsaan, Kementerian Kesihatan Malaysia)*

Note:

The availability of treatment or intervention should also be considered in order to decide if a disease should be listed. However, it is not a mandatory criterion.

The purpose of having the list is: -

1. To support the policy planning and development of a holistic management of rare diseases in Malaysia under the National Framework for Rare Disease in Malaysia.
2. To facilitate and ensure harmonization in the designation and registration of orphan medicines.

The list will be revised periodically to keep the list current and in line with newly acquired knowledge, real-world evidence, and new treatment options.

For further information, please contact:

Medical Development Division
Obstetrics & Gynaecology, Paediatrics Services Unit
Ministry of Health Malaysia

MALAYSIAN RARE DISEASE LIST

* The list was updated as of August 2020 and will be updated from time to time by the National Rare Disease Committee, Ministry of Health Malaysia.

Source: Medical Development Division, Ministry of Health, Malaysia

1. RARE INHERITED METABOLIC DISEASES

No	Disease name	ICD 10 code
1.1	Urea cycle disorders	E72.2
1.2	Glutaric aciduria type 1	E72.3
1.3	Propionic aciduria	E71.1
1.4	Methylmalonic aciduria	E71.1
1.5	Isovaleric aciduria	E71.1
1.6	3-Hydroxy-3-methylglutaric aciduria (HMG-CoA lyase deficiency)	E71.1
1.7	Maple syrup urine disease	E71.0
1.8	Phenylalanine hydroxylase deficiency (Classical phenylketonuria)	E71.0
1.9	Tyrosinaemia type I	E70.2
1.10	Alkaptonuria	E70.2
1.11	Cystathionine beta-synthase deficiency (classical homocystinuria)	E72.1
1.12	Isolated sulfite oxidase deficiency	E72.1
1.13	Phosphoglycerate dehydrogenase deficiency (serine deficiency disorders)	E72.8
1.14	Nonketotic hyperglycinaemia	E72.5
1.15	Ornithine aminotransferase deficiency (Gyrate atrophy of retina and choroidae)	E72.4
1.16	Lysinuric protein intolerance	E72.3
1.17	Cystinuria	E72.0
1.18	Classical galactosaemia	E74.2
1.19	Hereditary fructose intolerance	E74.1
1.20	Transaldolase deficiency	E74.9
1.21	Glycerol kinase deficiency	E74.9
1.22	Glucose transporter I deficiency	E74.9
1.23	Glucose-galactose malabsorption	E74.3
1.24	Fructose-1,6-bisphosphatase deficiency	E74.1
1.25	Pyruvate carboxylase deficiency	E74.4
1.26	Glycogen storage disease type 1a	E74.0

No	Disease name	ICD 10 code
1.27	Glycogen storage disease type 1b	E74.0
1.28	Glycogen storage disease type III	E74.0
1.29	Glycogen storage disease type IV	E74.0
1.30	Carnitine transporter deficiency	E71.3
1.31	Carnitine palmitoyltransferase I (CPTI) deficiency	E71.3
1.32	Carnitine acylcarnitine translocase deficiency	E71.3
1.33	Carnitine palmitoyltransferase II (CPTII) deficiency	E71.3
1.34	Very long - chain acyl CoA dehydrogenase deficiency	E71.3
1.35	Medium - chain acyl CoA dehydrogenase deficiency	E71.3
1.36	Mitochondrial trifunctional protein deficiency	E71.3
1.37	Multiple acyl-CoA dehydrogenase deficiency	E71.3
1.38	Succinyl-CoA:3-Oxoacid-CoA transferase (SCOT) deficiency	E79.8
1.39	Malonyl CoA decarboxylase deficiency	E79.8
1.40	Pyruvate dehydrogenase complex deficiency	E74.4
1.41	Kearns-Sayre syndrome	E88.8
1.42	Chronic Progressive External Ophthalmoplegia (CPEO)	E88.8
1.43	Mitochondrial Encephalopathy Lactic Acidosis and Stroke-like Episodes (MELAS)	E88.8
1.44	Myoclonic Epilepsy Associated with Ragged Red Fibres (MERRF)	E88.8
1.45	Neuropathy Ataxia and Retinitis Pigmentosa (NARP)	E88.8
1.46	Leber's Hereditary Optic Neuropathy (LHON)	E88.8
1.47	Leigh Syndrome	E88.8
1.48	Mitochondrial DNA Depletion Syndromes	E88.8
1.49	Primary mitochondrial disorders	E88.8
1.50	Primary Coenzyme Q10 deficiency	E88.8
1.51	Ethylmalonic encephalopathy (ETHE1)	E88.8
1.52	Creatine biosynthesis defect	E88.8
1.53	Adenylosuccinate lyase deficiency	E79.8
1.54	Adenosine deaminase deficiency	E79.8
1.55	Purine nucleoside phosphorylase [PNP] deficiency	D81.5
1.56	Deoxyguanosine kinase deficiency	E79.8
1.57	Lesch-Nyhan syndrome	E79.1
1.58	Thymidine phosphorylase deficiency [Mitochondrial neurogastrointestinal encephalomyopathy (MNGIE)]	E79.8
1.59	Thymidine kinase 2 deficiency	E79.8

No	Disease name	ICD 10 code
1.60	Aicardi-Goutières syndrome (AGS)	E88.8
1.61	Smith – Lemli – Opitz syndrome	Q87.1
1.62	X-linked dominant chondrodysplasia punctata 2 (Conradi-Hunermann syndrome)	Q77.3
1.63	Cerebrotendinous xanthomatosis	E75.5
1.64	Porphyria	E80.0
1.65	Congenital disorders of glycosylation	E74.4
1.66	MPS I, Hurler, Scheie disease	E76.0
1.67	MPS II, Hunter disease	E76.1
1.68	MPS IIIA, Sanfilippo A disease	E76.2
1.69	MPS IIIB, Sanfilippo B disease	E76.2
1.70	MPS IIIC, Sanfilippo C disease	E76.2
1.71	MPS IIID, Sanfilippo D disease	E76.2
1.72	MPS IVA, Morquio A disease	E76.2
1.73	MPS VI, Maroteaux-Lamy disease	E76.2
1.74	MPS VII, Sly disease	E76.2
1.75	Fucosidosis	E77.1
1.76	Sialidosis	E77.1
1.77	GM1 – gangliosidosis	E75.1
1.78	GM2 – gangliosidosis	E75.0
1.79	Gaucher disease	E75.2
1.80	Krabbe disease	E75.2
1.81	Metachromatic leukodystrophy	E75.2
1.82	Fabry disease	E75.2
1.83	Niemann-Pick disease type A or B	E75.2
1.84	Niemann-Pick disease type C	E75.2
1.85	CLN1, Santavuori-Haltia disease	E75.4
1.86	CLN2, Jansky-Bielschowsky disease	E75.4
1.87	CLN3, Batten Spielmeier-Vogt disease	E75.4
1.88	Neuronal ceroid lipofuscinoses type 6, CLN 6	E75.4
1.89	Neuronal ceroid lipofuscinoses type 7, CLN 7	E75.4
1.90	Cystinosis	E72.0
1.91	Mucopolipidosis II, I-cell disease	E77.0
1.92	Mucopolipidosis III, Pseudo-Hurler polydystrophy	E77.0

No	Disease name	ICD 10 code
1.93	Multiple sulphatase deficiency	E76.2
1.94	Wolman/cholesterol ester storage disease	E75.5
1.95	Pompe disease, GSD type II	E74.0
1.96	Galactosialidosis	E77.1
1.97	Pycnodysostosis	E88.8
1.98	Hermansky-Pudlak Syndrome	E70.3
1.99	Zellweger spectrum disorder	Q87.8
1.100	Rhizomelic chondrodysplasia punctata	Q77.3
1.101	X-linked adrenoleukodystrophy	E71.3
1.102	Peroxisomal fatty acid oxidation defects	E88.8
1.103	Refsum disease	G60.1
1.104	Primary hyperoxaluria type I	E88.8
1.105	Tyrosine hydroxylase deficiency	E70.8
1.106	Aromatic L-amino acid decarboxylase deficiency	E72.8
1.107	Succinic semialdehyde dehydrogenase deficiency	E72.2
1.108	GABA transaminase deficiency	E72.8
1.109	Guanosine 5 triphosphate cyclohydrolase I deficiency	E70.1
1.110	6-Pyruvoyl-tetrahydropterin synthase deficiency	E74.4
1.111	Sepiapterin reductase deficiency	E70.1
1.112	Methylenetetrahydrofolate reductase deficiency	E71.1
1.113	Disorder of cobalamin absorption due to intrinsic factor deficiency	D51.1
1.114	Defect in adenosylcobalamin synthesis-cbl A	E71.1
1.115	Defect in adenosylcobalamin synthesis-cbl B	E71.1
1.116	Disorders of intracellular cobalamin metabolism	E72.8
1.117	Biotinidase deficiency	D81.8
1.118	Holocarboxylase synthetase deficiency	D81.8
1.119	Pyridoxine-dependent seizures (antiquitin deficiency)	D88.8
1.120	Pyridoxal 5'-phosphate deficiency	E53.1
1.121	Molybdenum cofactor deficiency	E79.8
1.122	Menkes syndrome	E83.0
1.123	Fanconi-Bickel disease (GLUT-2 deficiency)	E74.0

2. RARE NEUROLOGICAL & NEUROMUSCULAR DISEASES

No	Disease name	ICD 10 code
2.1	Pelizaeus-Merzbacher disease	E75.2
2.2	Rett syndrome	F84.2
2.3	Huntington's disease	G10.0
2.4	Spinocerebellar ataxia	G11.0
2.5	Friedreich's ataxia	G11.1
2.6	Ataxia telangiectasia	G11.3
2.7	Hereditary spastic paraplegia	G11.4
2.8	Spinal muscular atrophy	G12.0
2.9	Amyotrophic lateral sclerosis (ALS)	G12.2
2.10	Multiple sclerosis	G35.0
2.11	Alexander disease	G37.8
2.12	Charcot-Marie-Tooth disease	G60.0
2.13	Congenital insensitivity to pain with anhidrosis (CIPA)	G60.9
2.14	Myotonia dystrophica	G71.1
2.15	Hypokalaemic periodic paralysis	G72.3
2.16	Joubert syndrome	Q04.9
2.17	Tuberous sclerosis	Q85.1
2.18	Neurofibromatosis type I	Q85.5
2.19	Neurofibromatosis type II	Q85.5
2.20	Acute necrotizing encephalopathy of childhood	G31.8
2.21	Alternating hemiplegia of childhood	G98
2.22	Ataxia with vitamin E deficiency	G11.1
2.23	Autoimmune encephalitis	G04.81
2.24	Autosomal dominant nocturnal frontal lobe epilepsy	G40.0
2.25	Dopa-responsive dystonia	G24.1
2.26	Bickerstaff's brainstem encephalitis	G61.0
2.27	Canavan disease	E75.2
2.28	Childhood absence epilepsy	G40.3
2.29	Childhood ataxia with diffuse central nervous system hypomyelination	E75.2
2.30	Chronic inflammatory demyelinating polyneuropathy	G61.8
2.31	Complex regional pain syndrome	G56.4
2.32	Congenital central hypoventilation syndrome	G47.3

No	Disease name	ICD 10 code
2.33	Dravet syndrome	G40.4
2.34	Early infantile epileptic encephalopathy	G40.3
2.35	Early myoclonic encephalopathy	G40.4
2.36	Epilepsy of infancy with migrating focal seizures	G40.8
2.37	Epilepsy with myoclonic absences	G40.4
2.38	Epilepsy with myoclonic-atonic seizures	G40.4
2.39	Epileptic encephalopathy with continuous spike-and-wave during slow sleep	F80.3
2.40	Episodic ataxia	G11.8
2.41	Febrile infection-related epilepsy syndrome	G40.5
2.42	Folinic acid-responsive seizures	G40.3
2.43	Hereditary sensory and autonomic neuropathy	G60.8
2.44	Hyperekplexia	G25.8
2.45	Hypomyelination with atrophy of basal ganglia and cerebellum	E75.2
2.46	Hypothalamic hamartomas with gelastic seizures	G40.5
2.47	Idiopathic acute transverse myelitis	G37.3
2.48	Idiopathic torsion dystonia	G24.1
2.49	Infantile spasms	G40.4
2.50	Juvenile absence epilepsy	G40.3
2.51	Juvenile myoclonic epilepsy	G40.3
2.52	Landau-Kleffner syndrome	F80.3
2.53	Lennox-Gastaut syndrome	G40.4
2.54	Limbic encephalitis with NMDA receptor antibodies	G13.1
2.55	Neurodegeneration with brain iron accumulation	G23.0
2.56	Neuromyelitis optica	G36.0
2.57	Ohtahara syndrome	G40.8
2.58	Opsoclonus-myoclonus-ataxia syndrome	G25.3
2.59	Primary dystonia	G24.1
2.60	Progressive myoclonic epilepsy	G40.3
2.61	Rasmussen syndrome	G04.8
2.62	Rapid-onset childhood obesity hypothalamic dysfunction hypoventilation- autonomic dysregulation (ROHHAD) syndrome	E66.2 E23.3 G90.8
2.63	Subacute sclerosing panencephalitis	A81.1
2.64	Subependymal giant cell astrocytoma	D43.2

No	Disease name	ICD 10 code
2.65	Benign essential blepharospasm	G24.5
2.66	Early onset generalized dystonia	G24.9
2.67	Focal dystonia	G24.9
2.68	Rippling muscle disease	G71.8
2.69	Narcolepsy	G47.4
2.70	Paraneoplastic neuromyopathy and neuropathy	G13.0
2.71	Myasthenia gravis	G70.0
2.72	Bethlem myopathy	G71.0
2.73	Central core disease	G71.2
2.74	Centronuclear myopathy	G71.2
2.75	Congenital muscular dystrophy	G71.2
2.76	Desmin-related myofibrillar myopathy	G71.8
2.77	Multiminicore myopathy	G71.2
2.78	Myotonia congenita	G71.1
2.79	Myotonic dystrophy	G71.1
2.80	Duchenne muscular dystrophy	G71.0
2.81	Nemaline Rod Myopathy	G71.2
2.82	Schwartz Jampel syndrome	G78.8
2.83	Facioscapulohumeral Muscular Dystrophy	G71.1
2.84	Myotubular Myopathy	G71.2
2.85	Becker Muscular Dystrophy	G71.0
2.86	Limb-girdle Muscular Dystrophy	G71.0
2.87	Congenital myasthenic syndrome	G70.2
2.88	Leukodystrophy	E75.2

3. RARE SKIN DISEASES

No	Disease name	ICD 10 code
3.1	Epidermolysis bullosa	Q81.0
3.2	Inherited ichthyosis	Q80.0
3.3	Ectodermal dysplasias	Q82.4
3.4	Dyskeratosis congenita	Q82.8
3.5	Incontinentia pigmenti	Q82.3
3.6	Netherton syndrome	Q82.8

No	Disease name	ICD 10 code
3.7	Xeroderma pigmentosum	Q82.1
3.8	Congenital generalized lipodystrophy	E88.1
3.9	Acrodermatitis enteropathica	E83.2
3.10	Albinism	E70.3
3.11	PTEN hamartoma tumour syndrome	E71.440
3.12	Klippel-Trenaunay syndrome	Q87.2
3.13	Keratitis-Ichthyosis-Deafness (KID) syndrome	Q80
3.14	Congenital hemidysplasia with ichthyosiform erythroderma and limb defects (CHILD syndrome)	Q77.3
3.15	Gorlin syndorome	Q85
3.16	Complex vascular malformation	D18.0
3.17	Autoimmune blistering disease	
	• Bullous pemphigoid	L12.0
	• Pemphigus vulgaris	L10.0
	• Pemphigus foliaceus	L10.2
	• Linear IgA bullous dermatosis	L13.8
	• Epidermolysis bullosa acquisita	L12.3
	• Dermatitis herpetiformis	L13.0
	• Paraneoplastic pemphigus	L10
• Cicatricial pemphigoid	H13.3	
3.18	H syndrome	Q82.9
3.19	Generalized pustular psoriasis	L40.1
3.20	Hailey-hailey disease/Benign familial pemphigus	L11.8
3.21	Darier's disease	L11.8
3.22	Pyoderma gangrenosum	L08.0
3.23	Porokeratosis	Q82.8

4. RARE ENDOCRINE DISEASES

No	Disease name	ICD 10 code
4.1	Pseudohypoparathyroidism	E20.1
4.2	hypophosphatemic rickets	E83.3
4.3	Laron syndrome (Laron Dwarfism)	E34.3
4.4	Bardet-Biedl syndrome	Q87.8
4.5	Congenital hyperinsulinism	E16.1
4.6	Congenital adrenal hypoplasia	Q89.1

No	Disease name	ICD 10 code
4.7	Congenital adrenal hyperplasia	E25.0
4.8	Kallmann syndrome	E23.0
4.9	Adrenocortical carcinoma	C74.9
4.10	Craniopharyngioma	D44.4
4.11	Disorders of sex development	Q56.4
4.12	Neonatal diabetes	P70.2
4.13	Pallister-Hall syndrome	D33.0
4.14	Cushing disease/syndrome	E24.0
4.15	Acromegaly/gigantism	E22.0
4.16	Neuroendocrine neoplasm	C7A.1
4.17	Pheochromocytoma	C74.1
4.18	Paraganglioma	D44.7
4.19	Familial endocrine tumour syndrome (multiple endocrine neoplasia)	E31.2
4.20	Central diabetes insipidus	E23.2
4.21	Nephrogenic diabetes insipidus	N25.1
4.22	Primary adrenocortical insufficiency	E27.1
4.23	Polyostotic fibrous dysplasia (McCune-Albright syndrome)	Q78.1
4.24	Alstrom syndrome	E34.8
4.25	Primary pituitary hypophysitis	E23.6
4.26	ROHHAD Rapid-onset Obesity with Hypothalamic dysfunction Hypoventilation Autonomic Dysregulation	E66.2 E23.3 G90.8
4.27	Vitamin D dependent rickets type 2	E83.3
4.28	Pseudohypoaldosteronism type 1	N25.8
4.29	Wolfram syndrome (DIDMOAD)	E34.8
4.30	Familial hypercholesterolemia (severe homozygous/autosomal recessive)	E78.01
4.31	Congenital generalised lipodystrophy (Berardinelli-Seip syndrome)	E88.1
4.32	Donohue syndrome (Leprechaunism)	E34.8

5. RARE DISEASES AFFECTING BONE, CARTILAGE AND CONNECTIVE TISSUE

No	Disease name	ICD 10 code
5.1	Hypophosphatasia	E83.39
5.2	Achondroplasia	Q77.4
5.3	Osteogenesis imperfecta	Q78.0
5.4	Ehlers-Danlos syndrome	Q79.6
5.5	Osteochondrodysplasias (Primary bone dysplasias)	Q78
5.6	Osteopetrosis	Q78.2
5.7	Syndromic craniosynostosis	Q75.0

6. RARE RHEUMATOLOGICAL DISEASES

No	Disease name	ICD 10 code
6.1	Acquired purpura fulminans	D65
6.2	Kawasaki disease	M30.3
6.3	Pigmented villonodular synovitis	M12.2
6.4	Osteonecrosis	M87
6.5	Polymyalgia Rheumatica	M35.3
6.6	Felty Syndrome	M05.0
6.7	Dermatomyositis	M33.0
6.8	Polymyositis	M33.2
6.9	Inflammatory Inclusion body myositis	M60.8
6.10	Primary Sjogren syndrome	M35.0
6.11	Acquired thrombotic thrombocytopenic purpura	M31.1
6.12	Juvenile Idiopathic Arthritis	M08.0
6.13	Idiopathic juvenile osteoporosis	M81.5
6.14	SAPHO syndrome	M86.3
6.15	Adult-onset Stills disease	M06.1
6.16	Diffuse Systemic sclerosis	M34.0
6.17	Systemic polyarteritis nodosa	M30.0
6.18	Autoimmune necrotising myositis	G72.4
6.19	Paediatric systemic lupus erythematosus	M32.0
6.20	Pulmonary arterial hypertension associated with connective tissue disease	I27.2
6.21	Pyogenic arthritis-pyoderma gangrenosum-acne syndrome	M04.8

No	Disease name	ICD 10 code
6.22	Pauci immune glomerulonephritis	N05.7
6.23	Pediatric Castleman disease	D36.0
6.24	PFAPA (Periodic fever - aphthous stomatitis-pharyngitis - adenopathy) syndrome	E85.0
6.25	Henoch Schonlein Purpura	D69.0
6.26	Takayasu arteritis	M31.4
6.27	Beh et disease	M35.2
6.28	Hyperimmunoglobulin D with periodic fever	E85.0
6.29	Eosinophilic granulomatosis with polyangiitis*	M30.1
6.30	Microscopic polyangiitis	M31.7
6.31	Primary vasculitis of central nervous system	I67.7
6.32	Mixed connective tissue disease	M35.1
6.33	NLRP-12 associated hereditary periodic fever syndrome	E85.0
6.34	Catastrophic antiphospholipid syndrome	D68.6
6.35	Blau syndrome	M04.8
6.36	Sarcoidosis	D86.0
6.37	Chronic nonbacterial osteomyelitis (Chronic recurrent multifocal osteomyelitis)	M86.3
6.38	Majeed syndrome	M04.8
6.39	Granulomatosis with polyangiitis	M31.3
6.40	IgG4 related disease	M35.0 D89.89
6.41	Giant cell arteritis	M31.5
6.42	Ankylosing spondylitis	M45.0-9
6.43	Rheumatoid arthritis	M05
6.44	Systemic lupus erythematosus	M32
6.45	Raynaud's syndrome	I73
6.46	Psoriatic arthritis	L40.5

7. RARE HEMATOLOGICAL DISEASES

No	Disease name	ICD 10 code
7.1	Atypical haemolytic uremic syndrome (aHUS)	D58.8
7.2	Paroxysmal nocturnal haemoglobinuria (PNH)	D59.5
7.3	Haemophilia with inhibitor	D66

No	Disease name	ICD 10 code
7.4	Primary thrombophilia	D68.5
7.5	Glanzmann thrombasthenia	D69.1
7.6	Erdheim–Chester disease (non-LCH)	D76.3
7.7	Multicentric Castleman Disease	D36.0
7.8	Wiskot–Aldrich syndrome	D82.0
7.9	Acquired haemophilia	D68.4
7.10	Autosomal recessive thrombotic thrombocytopenic purpura	M31.1
7.11	Acquired thrombotic thrombocytopenic purpura	M31.1
7.12	Langerhans cell histiocytosis (LCH)	C96.0
7.13	Haemophagocytic lymphohistiocytosis (HLH)	D76.1
7.14	Severe aplastic anaemia (SAA)	D61.9
7.15	POEMS syndrome	D47.7
7.16	AL amyloidosis	E85.9
7.17	Cold haemagglutinin disease (CHAD)	D59.1
7.18	Diamond Blackfan Anemia (DBA)	D61.01

8. RARE IMMUNOLOGICAL DISEASES

No	Disease name	ICD 10 code
8.1	Functional Disorders of Polymorphonuclear Neutrophils	D71
8.2	Immunodeficiency with Predominantly Antibody Defects	D80
8.3	Combined Immunodeficiencies	D81
8.4	Immunodeficiency associated with other major defects	D82
8.5	Common Variable Immunodeficiency	D83
8.6	Other Immunodeficiencies	D84
8.7	Other Disorders Involving the Immune Mechanism, not elsewhere classified	D89

9. RARE PULMONARY DISORDERS

No	Disease name	ICD 10 code
9.1	Cystic fibrosis	E84.0
9.2	Primary ciliary dyskinesia	Q34.8
9.3	Idiopathic pulmonary hemosiderosis	E83.1

No	Disease name	ICD 10 code
9.4	Congenital aerodigestive disease	J39.8
9.5	Primary interstitial lung disease	J84.9

10. RARE CARDIAC DISEASE

No	Disease name	ICD 10 code
10.1	Brugada syndrome	I49.8
10.2	Primary cardiomyopathy	I42.9

11. RARE RENAL DISEASES

No	Disease name	ICD 10 code
11.1	Autosomal recessive polycystic kidney disease	Q61.1
11.2	Autosomal dominant polycystic kidney disease	Q61.2
11.3	Alport Syndrome	Q87.8
11.4	Anti-glomerular basement membrane disease (Goodpasture)	N08.8
11.5	Atypical Haemolytic Uraemic Syndrome	D58
11.6	Autosomal recessive distal renal tubular acidosis	N25.8
11.7	Autosomal recessive proximal renal tubular acidosis	N25.8
11.8	Autosomal recessive polycystic kidney disease	Q61.1
11.9	(ARPKD) Batters syndrome	
11.10	Bardet-Biedl syndrome	Q87.8
11.11	Bartter syndrome	E26.8
11.12	BK-Virus Nephropathy	B97.89
11.13	Congenital nephropatic syndrome Finnish type	N04.8
11.14	Cystinosis	E72.0
11.15	Dense deposit disease (DDD)	N04.6
11.16	Denys-Drash Syndrome	N04.1
11.17	Dent Disease	N25.8
11.18	Exstrophy of bladder	Q64.1
11.19	Fabry disease	E75.2
11.20	Fanconi Anaemia	D61.0
11.21	Fanconi syndrome	E72.0
11.22	Familial hypomagnesaemia with hypercalciuria and nephrocalciuria	E83.4

No	Disease name	ICD 10 code
11.23	Focal segmental glomerulosclerosis	N04.1
11.24	Frasier syndrome	N04.1
11.25	Giant vessel / Giant cell arteritis	M31.6
11.26	Gitelman syndrome	N15.8
11.27	Hypophosphatemic rickets (X-linked)	E83.3
11.28	IgA vasculitis (Henoch Schonlein)	D69.0
11.29	Large vessel vasculitis	D69.0
11.30	Lesch Nyhan Syndrome	E79.1
11.31	Liddle syndrome	I15.1
11.32	Lowe's syndrome	E72.0
11.34	Medium vessel vasculitis	D69.0
11.35	Membranous nephropathy	N04.2
11.36	Membranoproliferative glomerulonephritis	N00.5
11.37	Microscopic polyangiitis	M31.7
11.38	Minimal change nephropathy	N04.0
11.39	Multicystic dysplastic kidneys (bilateral)	Q61.4
11.40	Nail patella syndrome	Q87.2
11.41	Nephropathic cystinosis	E72.0
11.42	Nephrogenic diabetes insipidus	N25.1
11.43	Nephrogenic syndrome of inappropriate antidiuresis	E22.2
11.44	Nephronophthisis	Q61.5
11.45	Primary hyperoxaluria	E74.8
11.46	Prune belly syndrome	Q79.4
11.47	Pure red cell aplasia	D60.0
11.48	Renal agenesis	Q60.0 – Q60.2
11.49	Renal coloboma syndrome	Q60.4
11.50	Shiga Toxin associated hemolytic uremic syndrome	D58.8
11.51	Small vessel vasculitis (ANCA associated)	I77.6
11.52	Steroid resistant nephrotic syndrome	N04.9
11.53	Steroid sensitive nephrotic syndrome	N04.9
11.54	Takayasu arteritis	M31.4
11.55	Tuberous sclerosis	Q85.1
11.56	WAGR (Wilm' tumour, Anirida, genitourinary anomalies and mental retardation)	Q87.8

12. RARE GASTROINTESTINAL AND HEPATIC DISEASES

No	Disease name	ICD 10 code
12.1	Progressive familial intrahepatic cholestasis	K76.8
12.2	α 1-Antitrypsin deficiency	E88.01
12.3	Congenital bile acid synthesis defect	E78.70
12.4	Wilson's disease	E83.0
12.5	Hereditary haemochromatosis	E83.1

13. RARE MALFORMATIONS, DEVELOPMENTAL ANOMALIES AND GENETIC SYNDROME

No	Disease name	ICD 10 code
13.1	Alagille syndrome	Q44.7
13.2	Beckwith Wiedemann syndrome	Q87.3
13.3	Marfan syndrome	Q87.4
13.4	Russel silver syndrome	Q87.1
13.5	Mowat-Wilson syndrome	Q43.1
13.6	Noonan syndrome	Q87.1
13.7	Cornelia de Lange syndrome	Q87.1
13.8	Kabuki syndrome	Q87.0
13.9	Sotos syndrome	Q87.3
13.10	Treacher collins syndrome	Q75.4
13.11	Turner syndrome	Q96.0
13.12	Fragile X syndrome	Q99.2
13.13	Cardiofaciocutaneous syndrome	Q87.8
13.14	Angelman syndrome	Q93.5
13.15	DiGeorge syndrome (22q11.2 deletion syndrome)	D82.1
13.16	Prader-Willi syndrome	Q87.1
13.17	WAGR syndrome (Wilms' tumor-Aniridia-Genitourinary anomalies-mental retardation)	Q87.8
13.18	Miller-Dieker syndrome	Q04.3
13.19	Rubinstein-Taybi syndrome	Q87.2
13.20	Williams syndrome	Q93.8
13.21	Von Hippel-Lindau disease	Q85.8
13.21	Cockayne syndrome	Q87.1

No	Disease name	ICD 10 code
13.23	Hutchinson-Gilford progeria syndrome	E34.8
13.24	Barth syndrome	E71.1
13.25	Costello syndrome	Q87.8
13.26	CHARGE syndrome	Q87.8
13.27	Coffin-Siris syndrome	Q87.1
13.28	Renal coloboma syndrome	Q60.4
13.29	Rare chromosomal abnormality with multiple malformations, physical and learning disabilities	Q99.9
13.30	Mayer-Rokitansky-Küster-Hauser syndrome	Q51.8

14. RARE INFECTIONS

No	Disease name	ICD 10 code
14.1	Herpes simplex viral encephalitis	B00.4
14.2	Extrapulmonary tuberculosis	A18.89

15. RARE CANCER

No	Disease name	ICD 10 code
15.1	Dermatofibrosarcoma protuberans	C49.9
15.2	Advanced melanoma	C43.9
15.3	Giant cell tumour of bone	D48.0
15.4	Malignant mesothelioma	C45.0
15.5	Iodine refractory thyroid cancer	C73
15.6	Oligodendroglioma	C71.9
15.7	Medulloblastoma	C71.6
	ADRENAL GLAND	
15.8	Adrenal Gland Carcinoma	C74.9
15.9	Adrenal cortical carcinoma	C74.0
15.10	Ganglioneuroblastoma	C74.9
15.11	Giant cell sarcoma (except of bone)	C74.9
15.12	Large cell neuroendocrine carcinoma	C74.9
15.13	Liposarcoma	C74.9
15.14	Lymphoma	C74.9

No	Disease name	ICD 10 code
15.15	Neuroblastoma	C74.9
15.16	Pheochromocytoma	C74.9
15.17	Primitive neuroectodermal tumor	C74.9
	ANUS	
15.18	Carcinoma of the anus	C21.0
15.19	Melanoma of the anus	C21.0
15.20	Lymphoma	C21.0
15.21	Neuroendocrine carcinoma	C21.0
15.22	Sarcoma	C21.0
15.23	Plasmacytoma	C21.0
	BILE DUCT	
15.24	Carcinoid tumor of the bile duct	C24.0
15.25	Klatskin tumor	C24.0
15.26	Lymphoma	C24.0
	BLADDER	
15.27	Sarcoma of the bladder	C67.9
15.28	Lymphoma	C67.9
	BONE	
15.29	All bone tumours	C41.9
	BRAIN	
15.30	Oligodendroglioma	C71.9
15.31	Ependymoma	C71.9
15.32	Lymphoma (any other terms)	C71.9
15.33	Medulloblastoma	C71.9
15.34	Intracranial Germ Cell Tumor	C71.9
15.35	Plasmacytoma	C71.9
	BREAST	
15.36	Lymphoma	C50.9
15.37	Carcinoid tumor	C50.9
15.38	Sarcoma	C50.9
15.39	Phylloides tumor	C50.9
	BRONCHUS / LUNG	
15.40	Adenoid cystic carcinoma	C34.9
15.41	Carcinoid tumor	C34.9

No	Disease name	ICD 10 code
15.42	Lymphoma	C34.9
15.43	Giant cell tumor	C34.9
15.44	Mesothelioma	C34.9

16. RARE EYE DISEASES

No	Disease name	ICD 10 code
16.1	Retinitis pigmentosa	H35.5
16.2	Familial exudative vitreoretinopathy	H35.0
16.3	Norrie disease	H35.5

APPENDIX B: ORPHAN DRUG DESIGNATION APPLICATION FORM

BAHAGIAN REGULATORI FARMASI NEGARA (NPRA) ORPHAN MEDICINE DESIGNATION APPLICATION FORM		
1. Date of Application:		
2. Information of Applicant (Product Registration Holder, PRH)		
Name of company:	Name of Contact Person:	
Address:	Tel No:	Fax No:
	E-mail address:	
3. Product Information		
Product Name:	Strength:	ATC Code:
Pharmaceutical Form:	Route of administration:	
Active Ingredient:	Strength:	
Manufacturer name and address:		
Worldwide regulatory status:		
Worldwide orphan medicine designation status:		
4. Proposed Rare Disease and Condition		
Proposed Indication related to the Rare Disease:		
<input type="checkbox"/> Treatment	<input type="checkbox"/> Prevention	<input type="checkbox"/> Diagnosis
Brief Description of Rare Disease:		
Current available method in treating/ preventing/ diagnosing the rare disease:		

4. Proposed Rare Disease and Condition (continued)

Justification for this product to be designated as orphan medicine:

Brief description of the product (details on active ingredient(s), medicines type/class, structure, physical-chemical properties):

Mechanism of action explaining how the product works in relevant disease/condition:

5. Scientific rationale for the orphan medicine use (the scientific rationale should support a medical plausible basis for the orphan medicine to be effective in treating disease/condition)

Please briefly describe the scientific evidence to support safety and efficacy of this product to treat/prevent/diagnose the proposed indication related to the rare disease:

Tabulated pre-clinical trial and clinical studies(Please enclose together with this form):

A brief safety update report:

6. Declaration of Applicant

- i) I hereby declare that all the information and attachment(s) provided are true.
- ii) I am fully aware of the consequences of rejection of this application if this form is incomplete.

.....
Name:

Company Stamp:

APPENDIX C: PERMOHONAN MEMPEROLEH DAN MENGGUNAKAN UBAT YANG MEMERLUKAN KELULUSAN KHAS (FOR MINISTRY OF HEALTH FACILITIES)

BPF/103-KPK01 (Pindaan 4.0)

PERMOHONAN MEMPEROLEHI & MENGGUNAKAN UBAT YANG MEMERLUKAN KELULUSAN KHAS KETUA PENGARAH KESIHATAN MALAYSIA / PENGARAH KANAN PERKHIDMATAN FARMASI

NO. SIRI**Hospital:****Bhg. Perkhidmatan Farmasi:****PERHATIAN:** Permohonan yang tidak lengkap **TIDAK** akan diproses.

1	Hospital yang memohon:	5	Nama ubat / keluaran dimohon (nama generik, kekuatan dan bentuk dosej):
2	Negeri:	6	Status pendaftaran Pihak Berkuasa Kawalan Dadah (PBKD: http://npra.moh.gov.my/)
3	Nama pesakit: Jantina: <input type="checkbox"/> Lelaki <input type="checkbox"/> Perempuan No. Kad Pengenalan pesakit: Umur pesakit: ____ tahun ____ bulan Berat: _____ kg Wad/Klinik: Diagnosis:	6.1 <input type="checkbox"/> a) Berdaftar dan tidak tersenarai dalam FUKKM: MAL <i>Indikasi seperti diluluskan oleh PBKD:</i> <input type="checkbox"/> Ya <input type="checkbox"/> Tidak* [Off-label PBKD] <input type="checkbox"/> b) Berdaftar dan tersenarai dalam FUKKM <i>Indikasi seperti tersenarai dalam FUKKM</i> <input type="checkbox"/> Tidak* [Off-label FUKKM] <input type="checkbox"/> Tidak* [Off-label PBKD] 6.2 <input type="checkbox"/> Tidak berdaftar tetapi tersenarai dalam FUKKM <i>Indikasi seperti tersenarai dalam FUKKM</i> <input type="checkbox"/> Ya <input type="checkbox"/> Tidak* 6.3 <input type="checkbox"/> Tidak berdaftar dan tiada dalam FUKKM* (perlu dinyatakan) Nama pengilang: Nama pengimport:	* Pemohon bertanggungjawab sepenuhnya terhadap penggunaan ubat tidak berdaftar dan di luar indikasi PBKD/FUKKM
4	Jenis permohonan: <input type="checkbox"/> Baru <input type="checkbox"/> Ulangan (No. siri kelulusan:.....)	7	Dos / regimen rawatan: Jangkamasa rawatan: 8 Kuantiti dimohon (maksimum 12 bulan): *Kos seunit: RM *Jumlah kos: RM (*sila sertakan sebutharga)

A	INDIKASI / TUJUAN RAWATAN BAGI UBAT / KELUARAN YANG DIMOHON		
B	RINGKASAN SEJARAH RAWATAN PESAKIT DAN JUSTIFIKASI PERMOHONAN		
C	UBAT / KELUARAN ALTERNATIF SEDIA ADA DALAM FUKKM BAGI INDIKASI YANG DIMOHON YANG TELAH DIGUNAKAN		
	UBAT / KELUARAN	TEMPOH PENGGUNAAN	SEBAB-SEBAB TIDAK DAPAT DIGUNAKAN/ DITERUSKAN
1			
2			
3			
D	PAKAR YANG MEMOHON		
	Ulasan:	Tandatangan, nama & cop: Tarikh:	
E	KETUA JABATAN		
	Ulasan:	Tandatangan, nama & cop: Tarikh:	
F	KETUA PEGAWAI FARMASI		
	Ulasan:	Peruntukan yang diperlukan: RM Peruntukan sedia ada: RM Tandatangan, nama & cop: Tarikh:	
G	PENGESAHAN PENGARAH HOSPITAL		
	Ulasan (sekiranya ada):	Tandatangan, nama & cop: Tarikh:	
H	PENGESAHAN PENERUSI JKK UBAT-UBATAN KKM (ruangan ini untuk kegunaan sekretariat)		
	Ulasan: <input type="checkbox"/> SOKONG <input type="checkbox"/> TIDAK SOKONG	Tandatangan, nama & cop: Tarikh:	

Source:

<https://www.pharmacy.gov.my/v2/sites/default/files/document-upload/borang-pesakit-updated-disember-2016-editable.pdf>

*Please refer updated version (if applicable)

APPENDIX D: APPLICATION TO IMPORT/ MANUFACTURE UNREGISTERED PRODUCTS FOR THE TREATMENT OF LIFE-THREATENING ILLNESSES (FOR PRIVATE/ NON-MINISTRY OF HEALTH INSTITUTIONS)

AKTA JUALAN DADAH 1952
PERATURAN-PERATURAN KAWALAN DADAH DAN KOSMETIK 1984
[PERATURAN 15(6)]

PERMOHONAN MENGIMPORT/MENGLILANG KELUARAN TIDAK BERDAFTAR BAGI
TUJUAN MERAWAT PENYAKIT YANG MENGANCAM NYAWA
(UNTUK INSTITUSI SWASTA/BUKAN DI BAWAH KKM)

No. siri/rujukan
Hosp/Syarikat:
Bhg. Perkhidmatan Farmasi:

PERHATIAN: Permohonan yang tidak lengkap **TIDAK** akan diproses.

1. Institusi yang memohon:	6	a. Nama (Keluaran & Bahan Aktif), Kekuatan (strength) & Bentuk Farmasetikal Keluaran dipohon: (Sila sertakan <i>clinical studies</i> dan maklumat berkenaan keluaran – Lihat Senarai Semak BPF/213-1) b. Pengilang: c. Syarikat Pengimport:
2. Jenis permohonan <input type="checkbox"/> Baru <input type="checkbox"/> Ulangan		
3. Nama & No KP/Pendaftaran pesakit (Sertakan lampiran jika ramai); atau nyatakan "Untuk Kegunaan Pesakit-Pesakit Institusi Ini" sekiranya untuk penyimpanan stok bagi kegunaan kecemasan		
4. a. Diagnosis / Indikasi: b. Ringkasan sejarah rawatan pesakit: c. Ubat berdaftar sedia ada (jika berkenaan): d. Tandakan sebab-sebab ubat berdaftar sedia ada tidak dapat digunakan: <input type="checkbox"/> Tidak berkesan <input type="checkbox"/> Kesan sampingan / kesan advers . Sila nyatakan:..... <input type="checkbox"/> Ubat berdaftar tidak dapat dibekalkan (sertakan surat maklumat daripada syarikat pemegang pendaftaran) <input type="checkbox"/> Lain-lain sebab. Nyatakan	7	a. Regimen dosej: b. Jangkamasa rawatan:
5. Pengesahan dari pakar perubatan yang memohon: (Sila nyatakan nombor pendaftaran sebagai Pengamal Perubatan – Lihat syarat di Lampiran BPF/213-1) (Nama, Tandatangan dan Cop) Tarikh: No, telefon / alamat emel untuk dihubungi:	8	a. Kuantiti dipohon (kuantiti penggunaan sehingga 1 tahun maksima – kuantiti per unit cthnya per vial/per tablet dsb): b. Harga kos seunit & jumlah kuantiti yang dipohon (Sila sertakan sebut harga jika ada):
	9	Pengesahan oleh Ahli Farmasi Institusi / Syarikat Pengimport Farmasetikal (jika institusi mempunyai Ahli Farmasi - Lihat syarat di Lampiran BPF/213-1): (Nama, Tandatangan dan Cop)
	10	Pengambilan Surat Pengecualian (Tanda yang berkenaan): <input type="checkbox"/> Secara serahan tangan di kaunter BPF, KKM kepada institusi. (Nama & no. untuk dihubungi/alamat emel): <input type="checkbox"/> Secara serahan tangan di kaunter BPF, KKM kepada wakil syarikat pengimport. (Nama & no. untuk dihubungi/alamat emel): <input type="checkbox"/> Pos berdaftar (sila ambil maklum mungkin mengambil masa dalam 1-2 minggu untuk diterima pemohon selepas mendapat kelulusan)

PERINGATAN: Sila rujuk LAMPIRAN BPF/213-1 bagi Panduan & Syarat-Syarat Mengisi Borang dan Senarai Semak BPF/213-1 Bagi Pemohon sebelum mengisi borang

Source:

<https://www.pharmacy.gov.my/v2/sites/default/files/document-upload/borang-bpf213-1-1-pindaan-2.pdf>

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