Ministry of Health & Medical Education
Rare Diseases Foundation of Iran
Tehran University of Medical Sciences

National Strategic Plan for Rare Diseases of Iran

In the Name of God, the Compassionate, the Merciful

National Strategic Plan for Rare Diseases of Iran

With the collaboration of the executive Departments,
Organizations and Research Centers

Collaborators:

Tehran University of Medical Sciences, Rare Diseases Foundation of Iran (RADOIR), Ministry of Health & Medical Education, Health Commission of the Islamic Consultative Assembly, Medical Committee of Rare Diseases Foundation of Iran, Shahid Beheshti University of Medical Sciences, Iran University of Medical Sciences, Secretariat of Supreme Council of Health & Food Safety, Non-governmental Rare Diseases Organizations & Communities

Statement by the Supreme Leader of Islamic Republic of Iran in a visit with MOH-Iran officials and Presidents of Universities of Medical Sciences of Iran dated Dec. 02, 1996

"The medical science should be significantly promoted to cover all patients healthcare needs. Increasing quantity in the field of medical sciences and promoting the scientific level cannot be separated from development issues. Therefore, the plans and programs by the government should be in a way that all of the people whether the rich or the poor, have access to the physician specialists and treatment referrals. To achieve this goal, development of low-cost medical services and facilitating the access to treatment is absolutely necessary".

Statement by Hojjatoleslam val-Moslemin Hassan Rouhani, President of the Islamic Republic of Iran

“Precision in diagnosis, enhancement of health services and reducing the people’s costs of living are the three principles of healthcare and treatment services. If we believe in the concept of development, firstly, we are to have healthy societies of any kind and the most essential base of development should be considered for human healthcare, science and technology”.

In the Name of God Almighty

The National Strategic Plan for Rare Diseases of Iran including an introduction and sixteen chapters approved by the Supreme Council of Health & Food Safety dated Dec. 26, 2020, is hereby notified to be strategically executed.

Hassan Rouhani
President of the Islamic Republic of Iran &
Chief of the Supreme Council of Health & Food Safety

In the name of God, the Compassionate, the Merciful

The Iranian nation has always taken efficient and abiding steps in promoting the country with immense empathy, excessive generosity and devotion within time periods. In line with serving the society, especially patients suffering from rare and difficult to cure diseases, the Ministry of Health & Medical Education has planned in different aspects and put the upstream national plans as agendum of its relevant departments.

The significance of prevention, diagnosis, treatment and social supports while serving the rare patients, should be necessarily prioritized in healthcare system performed by the experts at MOH-Iran within the several years of identifying rare diseases and prevention from the genetic disorders. Providing methods of novel processing of screening, diagnosis and treatment in the innovative medical fields has brought hopes to achieve successful recovery of various kinds of patients.

Providing methods of novel processing for screening, diagnosis and treatment of diseases in the innovative medical fields has brought hopes to achieve successful recovery of various patients of any types. Following the approved research methods applied in clinical trials assessment, they will be practiced in healthcare system. In Iran, new screening and treatment methods for rare diseases have been exploited and practiced along with the existing frameworks and defined in healthcare system instructions. Accordingly, the national strategic plan for rare diseases of Iran is counted as the most efficient step towards improving the status of the rare patients' diagnosis and treatment. It is hoped that the approved national strategic plan for rare disease of Iran acts as a turning point in prevention, treatment and promotion of living quality for the vulnerable rare patients.

Dr. Saeid Namaki
Minister of Health & Medical Education (MOH-Iran)

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Introduction

After the victory of the Islamic Revolution of Iran, the governmental authorities focus was mainly on the healthcare, treatment and developing new structures which resulted into considerable process in the existing statistics, accordingly. Therefore the changes in prevalence model of rare diseases, finding new treatment and diagnosis methods and also novel definitions and concepts as of health and welfare based on the sustainable development, require new approaches by the government and healthcare system in this regard.

Health is a universal right which should be accessible for all social classes, especially deprived and vulnerable stratum. For this reason and upon the citizenship rights, planning for promoting the health index, controlling and reducing social pathology in the lifestyle, education and employment of the rare patients are all counted as the governments' responsibilities.

Life threatening impacts of rare diseases are not limited to the rare patient and his/her family but the societies at national and international levels are exposed as well. Supplying and providing healthcare services are a part of duties by the governments through which continuous changes and challenges in the healthcare system and new detection methods of rare diseases, have become more complex and expensive due to the development of new sciences & technologies. Over multiple decades, most of the diseases have turned from communicable diseases to non-communicable and chronic ones. Rare diseases mainly refer to non-communicable diseases with genetic origin as one of the healthcare challenges in today's world left with no care and attention led to increasing population, disability and mortality which have brought an unpleasantness feature to the healthcare system and human societies in general.

Over the past decades, strategies and specific plans for the rare diseases were designed and developed in most countries and some international networks like ORPHANET collected data in regard with prevention, diagnosis and treatment methods.

According to the Sustainable Development Goals (SDG), the significance of planning for rare diseases, empowerment and high research potentiality of the scientific centers and also collaborating with healthcare experts, developing a comprehensive strategic plan for rare diseases by using the best of national resources and references, is an absolute necessity.

The National Strategic Plan for Rare Diseases of Iran is included with the definitions, generalities, macro strategic policies about rare diseases and the next phase of rare diseases strategic plan will refer in more details about the operational goals and solutions.

Global Status of Rare Diseases (Comparative Studies)

Since the most of the physicians have not encountered rare patients, their knowledge and information of the natural trend, therapeutic response and prognosis of such diseases are still incomplete. Obviously, these problems have limited the knowledge and understanding of rare diseases and have become a barrier for research and promising approaches, accordingly. Rare diseases registry is counted as a solution to prove their existence within the definite and strong evidence. Based on the concept, rare diseases registry system refers to the available organized or similar clinical data collection applied for the rare patients. Registry system has provided a unique opportunity to make progress and guideline for researching the clinical trials assessment and helps clinical decision makers. (1)

From the medical viewpoint and risk adjustment, practicing on diagnosis and treatment of rare diseases is not only an immediate need to guarantee healthcare and living rights of the suffering rare patients, but it is also an irresistible necessity to achieve unity, equity and justice which is of a high practical importance. (2)

Rare diseases have mostly intensified the side-effects and mortality and significantly have an impact on the rare patients' life style and have also imposed extra economic and social burden on the families and society. Although any types of the rare diseases affect a few individuals, based on the statistics there are more than 25 million US citizens and 30 million EU countries population suffering from rare diseases. On the other hand, some of the diseases defined as rare, may have also become endemic in a specific population and could have harmful effects on these societies. (3)

It is estimated that 5000 to 8000 rare diseases have been identified worldwide and impacted approximately 6 to 8 percent of the populations. Besides the rare diseases have many variants of etiology and clinical manifestations which mostly disabling with serious impacts on life expectancy as well as the physical and mental capabilities.

Yet, apart from the size and specifications of the population of a country, rare diseases have imposed an economic pressure and burden due to the high cost of healthcare and reduced efficiency levels of the society, they have caused an extra high pressure on the public budget of the country. (4)

Most of the rare patients experience difficulties and obstacles to access care and only less than 10% of them receive special treatment. Delay in diagnosis, limited access to the resources and lack of specific treatment methods, prevent the rare patients from receiving appropriate and on time care.

Delayed diagnosis may have deep impacts on the results of treatment. In more than 40% of rare patients, misdiagnosis will cause the delay in treatment. Even when the disease is diagnosed, most of them are not able to have access to the expertise

centers, coordinated cares, patient support systems and efficient treatments. Many of the rare diseases, have no certain treatment and the information about the progress of rare diseases is very limited. Therefore, researches on the natural trend of rare disease and basic pathophysiological mechanisms are necessary to discover the targeted drugs. (4)

Dealing with the challenges of rare patients, the society plays an important role by growing the voices of the rare patients to reach out developing the strategies to meet their needs. Almost as of 3.5 decades ago, raising awareness and supporting the rare disease patients in some countries such as the United States of America led to establish "National Organization for Rare Disorders" (NORD).(4)

Evaluating the impacts of rare diseases on the healthcare system has been very difficult. Firstly, because there is not a global integrated definition for rare diseases and their prevalence among different countries which ranges from 5 to 76 individuals in 100.000 (with a threshold of average 40 individuals in 100.000). Second of all, new diagnosis methods are appearing so rapidly that 100 types of new rare diseases identified per year (2010-2015). Thus few studies done about their burden and challenges on the healthcare system and mostly estimated through the national rare diseases or disabilities registry. These studies are helpful in defining the rare population characteristics but dependent on the reports by the specialist physicians and cannot reflect the complex data collection such as hospitalization or its duration. In a study in Hong Kong, rare patients had formed 1.5% of the population of the country while 4.3% of the same population was involved with their annual hospitalization cost and expenses. The average period of rare patients' hospitalization was 6.7 days and 1 day, respectively. Under 5 year groups had an average 8.7 days and 1 day hospitalization. Rare respiratory diseases had a long term and later on the ophthalmic rare diseases became the longest one. This inequality not only refers to the emergency costs of healthcare but it will also have considerable economic and social consequences for the rare patients, their family members and caregivers. (5)

Rare diseases have not been adequately studied and investigated in the world yet. Data registry systems in the form of biologic data banks have independent registry methods function (capacity of comprehensive collection of the clinical and epidemiologic data) and independent biological data banks registry methods (capability of OMICS research collaboration). Bio banks registry system method provides a unique, pragmatic, cost-effective and efficient solution for rare diseases researches. The relation of independent registry methods with rare diseases bio banks, provide appropriate resources for effective exploitation of the basic researches in clinical medicine. (6)

Although there are no consensus about definition, total number, estimated genetic origin and prevalence of rare diseases, there is no doubt that they are considered a

serious cognitive load to the society. Based on the national biostatistics report in the United States of America in 2016 (Haron 2018), congenital disorders, deformation and chromosomal disorders are considered 22.2% of mortality rate of neonatal while the rate of 20.8% of all mortalities belongs to breast feeding babies (main cause of death in this group) and for the age group 1 to 4 years old, it is 10.7%. In japan, the mortality rate percentage of breast feeding babies with congenital and chromosomal disorders is more (about 35.7%). (7)

In a report of 2005 at rare diseases conference held in Europe, the life expectancy of 323 rare patients were analyzed that 25.7% of them probably die before the age of 5 and more than 36.8% lost their life expectancy and only one third (37.5%) of them have normal life span.(7)

According to a study, 45.4% of patients suffering from genetic disorders were hospitalized 5 times or more, 26% of them 10 times or more and 12.8% of them, 20 times or more compared to total number of rare patients whom were sequentially 6.3, 5.8 and 10.7 times. In another study, not only the period of hospitalization of the rare patients with congenital and genetic diseases was more (average period of hospitalization 8.7 days compared to 5.7 days) than patients hospitalized for other reasons, but their expenses were also 2.8 times higher and the mortality rate of these patients were 4.5 times more as well. (7)

Mortality rate in the rare patients is considered 4.2% of all the lost years of life in public population which is more than 4 times of the lost years due to the infectious diseases and almost two times of the lost years belongs to diabetes. In 2017, the global sales of orphan drugs were about 125 billion dollars (with an annual growth of 11.3%) which comprises almost 15.9% of the wholesale of the prescribed non generic drugs. According to the same report, the average annual expense of each patient was 4.8 times comparing to the cost of orphan drugs (in comparison with non-orphan drugs). (7)

According to WHO policies, during the complicated processes of policy making, there must be some principles developed to identify multiple initiations for taking action to change and establish a strategic network of governmental beneficiaries and the civil society. It is also necessary that all the established networks have been integrated as a pivot operating platform along with the general compromising in the society to share data, research and required capacities in a supportive framework.(8)

World Health Organization has recommended two important admissions to manage the complex processes of rare diseases including approaches through “generality in the state or government” and “generality in the society”. These approaches should be compared and adjusted based on the special status and condition in every state or government. (8)

Sustainability of rare diseases healthcare systems with critical financial burdens, has raised more challenges for other groups of systems. There are lots of similarities between public health capacity, resilience in rare diseases and available mechanisms to ensure their sustainability including networking and empowering the rare patients and also redirecting healthcare towards the integrated care covered by the society and in-home caregiving. Networking and empowering the rare patients depend on the resistance and resilience of the society and healthcare systems; for example the fact how the individuals, groups and the majority of people in the society are seriously adjustable with challenges. Currently, the active role of rare patients representatives as a substantial part is known to accelerate cooperation and sustainability as well as their political role in raising awareness and seeking for national and international supports.(8)

The elements of an integrated national strategic plan for expediting the achievements of scientific and pragmatic research and development of rare diseases are:

- Active Collaboration of a vast group of public and private beneficiaries, such as governmental sector organizations, commercial companies, universities and academic centers, researchers and supportive groups.
- In time-Use of scientific and technological developments which can make the cost of rare diseases researches faster, easier and cheaper.
- Developing innovative strategies in sharing research and infrastructures for the optimal and efficient use of low financial resources, specialist experts, data, biological samples and the collaboration of rare patients in the researches.
- Making the best and efficient use as well as developing the experimental plans and analytical methods according to special challenges in line with the researches regarding small populations.
- Considering reasonable rewarding for innovations in the private sector and cautious use of the public resources for development of product when this method is counted as of the faster and low cost to meet the essential unmet needs of the rare patients.
- Monitoring adequate resources such as staff expertise in rare disease research and developing the medical equipment for public organizations responsible for supplying financial requirements of the biomedical researches as well as adopting the orphan drugs and medical equipment regulations.
- Mechanism of evaluating priorities of rare diseases researches, manufacturing of products, creating common and specific goals as well as their progress assessment to achieve. (9)

International Rare Disease Research Consortium (IRDiRC) has set up some goals for the years 2017 - 2027. IRDiRC members are obliged to meet three goals in line with the progress towards the vision of consortium for the upcoming decade.

The First Goal: If all the patients under clinical care with a suspicion of a rare disease identified and approved according to the medical references, should be diagnosed within a year and all who are not diagnosed by the time being, should enter a global and well organized route of diagnosis and research.

The Second Goal: There should be one thousand new treatment methods for rare diseases to be approved of which the most left without treatment, have to be under serious clinical investigation.

The Third Goal: New methods should be developed for evaluating the impacts of diagnosis and treatment on the rare diseases.(10)

During the recent years, many different countries have paid more attention to rare diseases such as the Chinese government which has merged rare diseases with its national healthcare strategies and programs. In 2018, the Chinese government officially released its first list of rare diseases included 121 types of rare diseases published to raise the public awareness about such types of diseases, to promote the capability frontline healthcare personnel working on the treatment of rare diseases, to develop the research rewards, to develop and facilitate the access to the orphan drugs. The effort had an increasing impact on rare diseases management potentiality in promoting the trends of diagnosis and treatment for the rare diseases in this country and also providing rights and resources related to the rare patients' healthcare. Classification of the rare diseases based on the common international standards will be followed by promoting the international collaboration of orphan drugs researches and policy making about the rare diseases as well.(11)

Rare Diseases Status in Iran

Unfortunately, there is no official and accurate definition of rare diseases by the Ministry of Health & Medical Education in our country; hence rare diseases are obsolete and also the rare patients sufferings have left behind.

Due to the lack of official definition as well as the comprehensive and integrated national rare patient registry system, there is not an accurate statistic of the existing rare patients in Iran. In the time being, the only electronic rare patients files registry systems in the country, has been running by the Rare Diseases Foundation of Iran (RADOIR). According to the acknowledgements and calls for registry, the rare patients and the medical specialists do the registry after data entries and uploading the required medical documentations through the Rare Patients Registry System (SABNA) and receiving confirmation by RADOIR medical committee including the specialist physicians and university professors, the registry process will be completed, accordingly. Based on the existing data in this system, there are 335 different types of rare diseases in the country and total number of approved rare patients' registries is 3140. Of course after 2 years of the establishment of SABNA rare patients registry system at which the volunteer rare patients register, it can be estimated that the statistics of rare diseases in the country, whether in terms of variety or quantity, is higher than the existing ones. The geographical distribution of rare diseases is determined and discussable into some extents. (Data and the diagrams annexed)

In its current programs specified for the screening, the important and prioritized rare diseases with the approach of research and offering services, the Ministry of Health & Medical Education has taken the necessary actions regarding rare diseases registry. One of the most valuable ones done specially for those with rare genetic origin at the Deputy health office of the Ministry of Health & Medical Education with the strategy of genetic assessment from birth to death as well as merging the aforesaid plan with various screening plans and health packages which will be applied permanently through implementation of the National Strategic Plan for Rare Diseases of Iran. In addition, the neonatal screening for treatable diseases with diagnosis and standard management in the framework of social genetic and merged with healthcare system, is one of the significant goals at the non-communicable diseases department of the Ministry of Health & Medical Education. The target diseases of "Neonatal Screening Program for the Congenital Metabolic Diseases" including 22 rare diseases of which 20 cases are diagnosable with MS-MS technology (including Amino Acids Disorders, Fatty Acid Disorders Chains & Organic Acidemia) and two of which (including Biotinidase and Galactosemia Deficiencies) via applying ELISA method.

The rare immunodeficiency diseases registry is another plan being performed by Immunodeficiency Diseases Research Center which has been able to register 3400 immunodeficiency rare patients so far.
With the aim of meeting the needs of rare metabolic patients, "Comprehensive Metabolic & Dietary Patients Needs System" is also available at MOH-Iran but not applied with special focus; therefore a lot of diseases are not covered and registered.
The point of necessity is that national registry system of a wide range of rare diseases requires a comprehensive nationwide instructions for better implementation of the diseases registry program with the assistive collaboration of all the universities of medical sciences and the affiliated health & treatment networks of each city for the next step.
According to a study conducted by Dr. Heydari and Colleagues in 2013, the cost of healthcare and treatment for Phenylketonuria is IRR 2,482,116 in case of the late diagnosis within a lifetime. Accordingly, the cost of treatment and care for each PKU case was estimated IRR 7,708.4. In the meantime, salary and allowances of the healthcare workers at the highest rate (46%) and hospitalization costs due to the seizures at the least rate (0.3%) were calculated.
The results of this study showed a considerable financial burden of PKU disease for the rare patients, their families and the government; therefore due to the high prevalence of this disease and in order to reduce the costs and consequences all over the country, the screening plan for this diseases should be developed and accessible for everyone to receive required services.
Based on the study conducted by Dr. Vameghi and her colleagues at the University of Social Welfare & Rehabilitation Sciences, among the factors of certain diagnosis on speech and language disorder in children, meaningful differences were detected, there were three factors related to the right consultation with the physicians, two factors considered for parents awareness and two others were directly relevant to child's screening program. According to the results, it was finally recommended to plan for raising public awareness, enhancing beliefs and attitudes of different classes of society and also providing proper education for raising up the knowledge as well as the approach of pediatricians and other pediatric diseases specialists. At the moment, the onset disorders and congenital diseases prevalence in Iran as a country with an average income, accounts for 3 to 5 percent of live births. In our country, the number of women who are still getting pregnant after the age of 35, is considerable while it is doubled in the societies with high incomes as per the assessments reports. Hence there are between 50,000 to 70,000 rare patients born in Iran.
In 2017, according to the statistics of institute for Health Metrics & Evaluation (IHME) in charge of the global burden of disease (GBD), the onset congenital

deficiencies were included 3.49% of the whole disability adjusted life years (DALY) in Iran, is higher than its global average which is about 2.44%. Based on the same statistics, the onset congenital disorders account for 1.32% of DALY and 1.79% of the whole mortality rate. Of course, if we add up the number of newborn disorders (some of which are related to the rare diseases) to these figures, the mortality and disability rate indexes will be higher.

As reported by the director of Pediatric Endocrinology Association (Dr. Rabbani), the late diagnosis of metabolic and rare hereditary diseases will involve the rare patients with neurological and mental side effects and will also leave a heavier burden on the government and healthcare system of Iran.

Most of the children suffering from metabolic diseases seem healthy and natural at birth but before the symptoms detected, it is possible to diagnose such diseases by screening test in order to prevent mental retardation and disabilities. Dr. Rabbani believes that the patterns of diseases tend to change into the non-communicable diseases and hereditary metabolic diseases which are generally caused by the familial marriages, are frequent problems and the rare patient has to take medicine all over his or her lifetime.

Unfortunately, these patients are generally treated emotionally while a comprehensive guideline should be developed for the rare patients treatment to improve their living quality. In Iran, about 1 in 600 newborns is suffering from hereditary metabolic diseases. These patients need different nutritional supplements from others and should stand on a special diet.

Diagram 1: Dispersal of Registered Rare Disease Patients in the Provinces of Iran

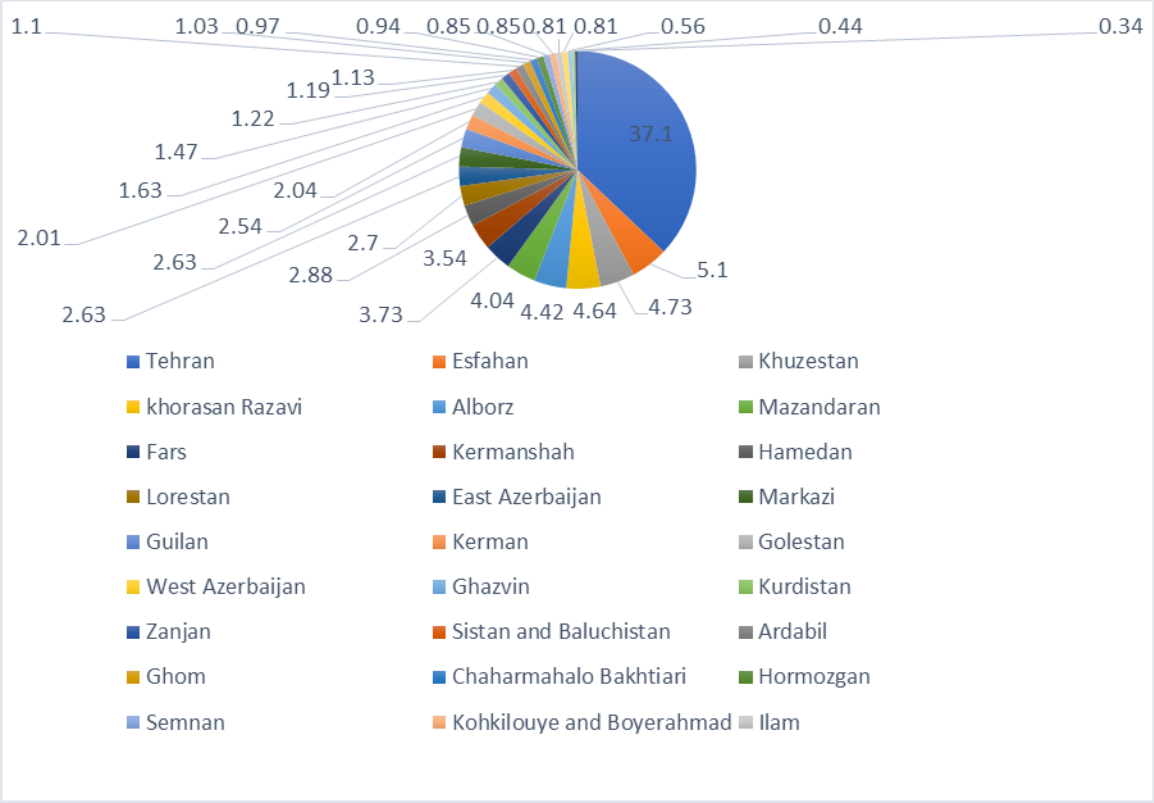


Diagram 2: The Decimal Percentage of Rare Diseases Types Registered in Iran

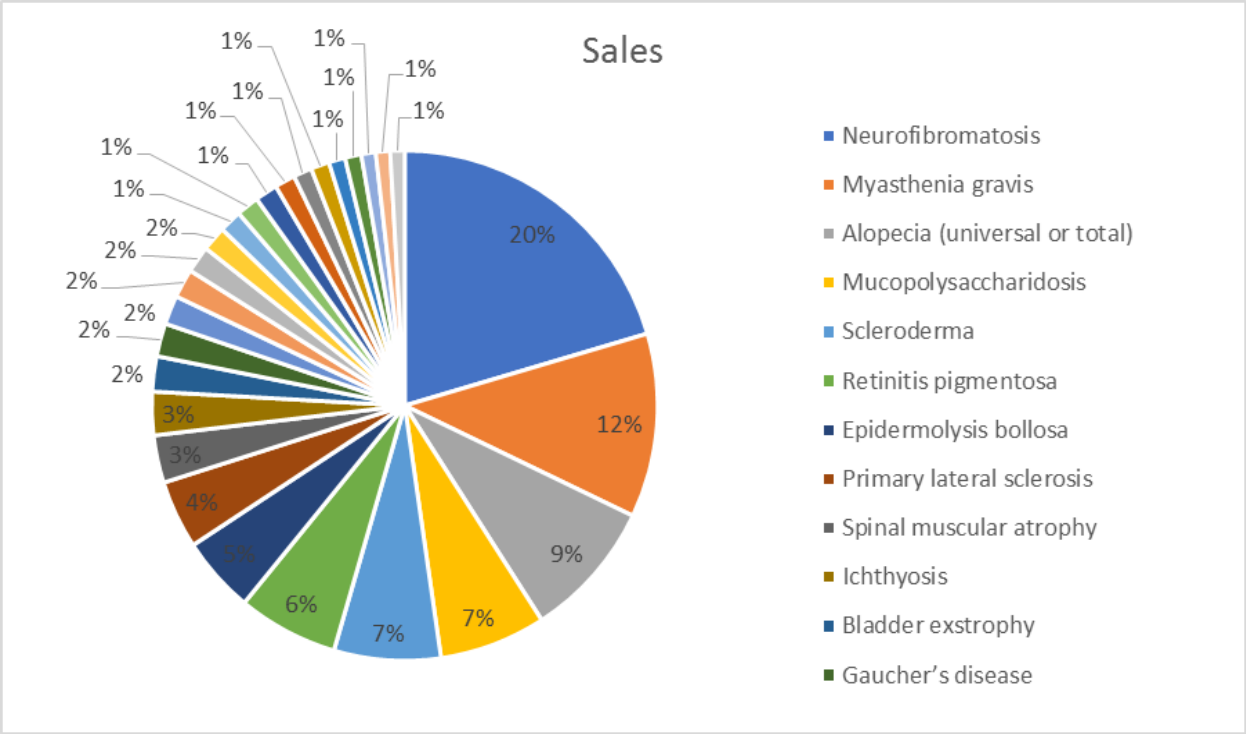
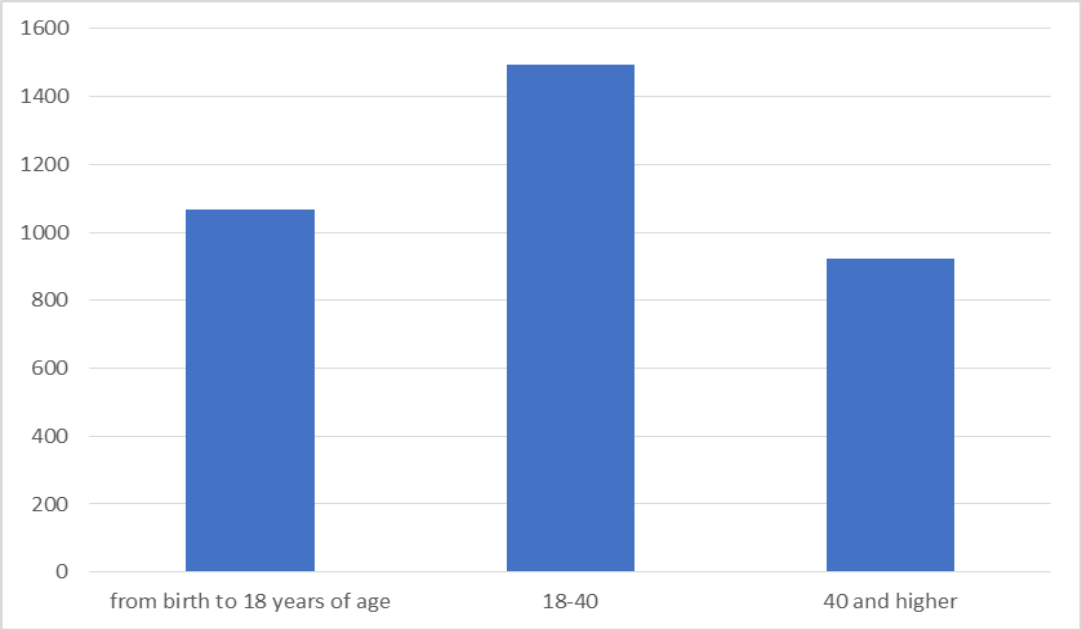


Table 1: Number of Registered Rare Patients in SABNA with ICD-10 Coding

Rare Disease	ICD-10	No.	Decimal
Neurofibromatosis	Q85	492	15.6%
Myasthenia Gravis	G70	280	8.9%
Alopecia (Universal or Total)	L63	210	6.7%
Mucopolysaccharidosis	E76.3	165	5.2%
Scleroderma	M34.9	164	5.2%
Retinitis Pigmentosa	H35.5	154	4.9%
Epidermolysis Bollosa	Q81.9	116	3.7%
Primary Lateral Sclerosis	G12.2	105	3.3%
Spinal Muscular Atrophy	G12.9	72	2.3%
Ichthyosis	Q82.83	67	2.1%
Bladder Exstrophy	Q64.1	54	1.7%
Gaucher's Disease	E75.21	51	1.6%
Achondroplasia	Q77.4	43	1.4%
Duchenne Muscular Dystrophy	G71	43	1.4%
Charcot-Marie-Tooth Disease	G60	42	1.3%
Phenylketonuria	E70.1	37	1.2%
Tuberous Sclerosis	Q85.1	35	1.1%
Cerebellum Atrophy	G31.9	34	1.1%
Cystinosis	E72.01	34	1.1%
Primary Immunodeficiency Disease	D83.9	33	1%
Osteogenesis imperfecta	Q78	27	0.9%
Mitochondrial Myopathy	G71.3	27	0.9%
Friederichs's Ataxia	G11.1	26	0.8%
Spinocerebral Ataxia	G11.8	25	0.8%
Limb Gridle Muscular Dystrophies	G71.01	21	0.7%
Marfan Syndrome	Q87.4	21	0.7%
Mayer Rokitansky Küster Syndrome	Q51.8	21	0.7%

Diagram 3: Dispersal of Rare Disease Patient Based on Age Spectrum Registered in Rare Patients Registry System (SABNA)



Age Spectrum of Rare Patuebts Registered	
Onset to 18 years of age	1066
18-40	1495
40 & higher	921

Number of Rare Patients by Gender	
Female	52%
Male	48%

Rendered Services to the Rare Patients of Iran

At the time being, the rare patients registered, identified and approved at Rare Disease Foundation of Iran (RADOIR) via its rare patients registry system (SABNA), have been provided with free or lower costs services based on the classification of rendering medical services protocol by RADOIR's medical committee. Another group of rare patients receive different services from the organizations such as State Welfare Organization, Imam Khomeini (God have mercy upon him) Relief Foundation, Insurance Organizations, Ministry of Education, NGOs and Public & Charity Care & Treatment Centers.
According to the existing data and statistics analysis (few and scattered) of rare diseases in the country, the notification of the National Strategic Plan for Rare Diseases of Iran seems quite necessary more than ever considering the essential approaches developed in IRAN'S 1404 (2025) VISION POLICY including healthcare and welfare provision and the healthcare system strategy based on prevention and education, diagnosis, medication & treatment and finally improving the life quality of the rare disease patients as well as setting out duties of beneficiaries for the important group of patients which will undoubtedly affect the future of healthcare system.

Developing Process of the National Strategic Plan For Rare Diseases of Iran

Considering the importance of rare diseases prevalence and incidence, there are numerous challenges as well as the financial, social, spiritual and human costs, the necessity of developing the national strategic plan for rare diseases of Iran and also a special rare patients guideline for these types of diseases, setting out duties of the relevant organizations, the significance of prompting the introduction and drawing the attention of different parts of Iran to make a precise and efficient policy, the Rare Diseases Foundation of Iran (RADOIR) was seriously determined to initiate and provide the required conditions in order to structuring and legitimizing the rare diseases issues which can reduce the expenses and the relevant challenges with such diseases in the society and healthcare system as well.

1- As per the suggestion and follow ups by the Late Dr. Ali Davoudian, the founder and CEO of rare Disease Foundation of Iran (RADOIR) as well as the immense effort of Mr. Yaser Davoudian, Chairman of RADOIR and also with the cooperation and support of members at the parliament of Islamic Republic of Iran, the authorities at the Ministry of Health & Medical

Education, especially the former Minister, Dr. Seyed Hassan Ghazizadeh Hashemi, received a notification letter dated Oct. 09, 2018 on developing a draft of the National Strategic Plan for Rare Diseases of Iran by RADOIR to be initiated upon the minister's approval.

2- A meeting invitation about developing the strategic plan for rare diseases was issued by NGOs general director and policy makers at the Ministry of Health & Medical Education and with the attendance of a number of MOH-Iran Officials, directors of rare diseases NGOs and the experts at Social Deputy office of the Ministry and specialized working group of developing the strategic plan was formed to have sessions every three months dated Dec. 03, 2018.
3- Following the necessary actions, a letter was sent to the Minister of Health & Medical Education, Dr. Seyed Hassan Ghazizadeh Hashemi emphasizing on the necessity of developing national strategic plan for rare diseases of Iran dated Dec. 18, 2018 as well as undertaking the financial costs & expenses and establishing the secretariat by the Rare Diseases Foundation of Iran (RADOIR), accordingly.
4- As the new Minister of Health & Medical Education, Dr. Namaki was appointed, a letter subjected "RADOIR assignment to develop the National Strategic Plan for Rare Diseases of Iran" was issued and sent by the Members of Parliament of the Islamic Republic of Iran dated Jan. 20, 2019 and upon the Minister's agreement, it was referred to the Deputy of Health and Treatment at MOH-Iran.
5- Investigating the subject, the Deputy of Treatment, Dr. Janbabaei agreed and with the cooperation of his colleagues, especially Dr. Shadnoush, Head of Management Center for Transplantation & Special Diseases and the president of Tehran University of Medical Sciences, Dr. Karimi were notified to cooperate with the RADOIR in developing the strategic plan through a letter dated March 16, 2019 for the attention of Dr. Sahraiean, Deputy of Research & Technology at TUMS.
6- Targeting the subject of developing the national strategic plan for rare diseases of Iran in the agenda of TUMS deputy of Research & Technology and RADOIR Deputy of Scientific, Educational & Research based on the precise and initiative studies as well as investigating the relevant issue as Up-Stream plan in the framework of developing the national plans, the strategic program and its operational principles were presented within several meeting sessions with TUMS.
7- At the council of strategic plans developments, professors and scientific board members from Tehran and Shahid Beheshti Universities of Medical

Sciences expertise in developing the national plans for healthcare systems were granted order of performance and notified dated May 28, 2019.

8- The first meeting of the council was held on June 16, 2019 regarding the revision of the strategic plan, finalizing time scheduling of the operational plan, methodology and definition of rare diseases, theoretical fundamentals, vision, macro objectives and strategies, national proceedings, proposed frameworks and executive commitments. Afterwards, developing the national strategic plan was assigned among the five different working groups:

- Collecting comments and viewpoints (supervised by Dr. Norouzi)
- Developing theoretical fundamentals of the national strategic plan and revising Up-Stream documentation of other countries and regions (supervised by Dr. Edraki)
- Rare diseases epidemiology (supervised by Dr. Shamshiri)
- Rare disease management (supervised by Dr. Salehpour & Dr. Abdi)
- Monitoring the International affairs and resources to conduct the comparative studies (supervised by Dr. Edraki)

9- Developing the national strategic plan for rare diseases of Iran was effected at the university council meetings and expert group sessions of various working groups. About fifty authorities & rare diseases experts were successfully interviewed by the first working group managed by Dr. Norouzi and their points of views were implemented and recorded to be applied for developing the national strategic plan for rare diseases of Iran. A survey was conducted among twenty rare diseases NGOs through questionnaire correspondence and the received comments were taken into careful consideration.

10- Working group for the international affairs & resources to conduct the comparative studies supervised by Dr. Edraki, were successfully inspected and translated the operational plans and relevant rare diseases references worldwide (about 15 countries), the international rare diseases authorities and communities were interviewed through questionnaire correspondence to be effected in developing the national strategic plan for rare diseases of Iran.

- Yann Le Cam – Chief Executive Officer of EURORDIS, Member of the Council of Rare Diseases International(RDI) and Vice-chairman of Rare Diseases NGOs Committee of the United Nations, New York
- Mr. Anders Olauson – Managing Director of Ågrenska Rare Diseases Foundation in Sweden, General Secretary of Rare Diseases Forum of Europe

and senior counsellor of Rare Diseases NGOs Committee of the United Nations, Sweden

- Dr. Francesco Pollini – Head of services department (National Plans Coordination Service at Center of Luxembourg (ESTERNE) - MOH-Belgium & Coordinating Manager of Rare Diseases National Strategic Plans, Belgium
- Mrs. Jimmy Harvey – CEO at International Coalition of Organizations Supporting Endocrine Patients (ICOSEP), Switzerland
- Mr. Jerald Lober – CEO at International Society for Neonatal Screening (ISNS), Switzerland & China

11- The opinions of national and international experts and officials were organized in a pamphlet collection and so far nine university council meetings and thirty-seven expert group sessions held, totally. Eventually, due to the immense effort and intensive cooperation of Tehran University of Medical Sciences (TUMS) and deputies of science, international affairs and treatment at Rare Diseases Foundation of Iran (RADOIR), the initial copy of the national plan was presented and approved by the members at the strategic council of developing the national strategic plan for rare diseases of Iran dated Oct. 05, 2019.

12- Pursuant to the letter dated Oct. 26, 2019, the drafted national strategic plan was sent by TUMS president enclosed with annexes to the deputy office of treatment at MOH-Iran including the national plan text, viewpoints pamphlet collection of the elite theoreticians, from the beginning/end of documented correspondences were sent to TUMS president by the deputy of scientific research at RADOIR.

13- Pursuant to the letter dated Oct. 26, 2019 by TUMS president to the deputy office of treatment at MOH-Iran in order to proceed the upcoming steps in the Ministry of Health & Medical Education as well as the Supreme Council of Health & Food Security to be attended by the minister of health, Dr. Namaki.

14- After several expert group meetings about the various levels of beneficiaries with the representatives of organizations and rare diseases communities, the necessary amendments were made and the national strategic plan was approved at the 3rd permanent commission of supreme council of alimentary health and security on Dec. 26, 2020 accompanied by all members, deputies or representatives of the ministries as well as the executive organs, held by the different ministries and executive departments and chaired by the Minister of Health & Medical Education.

Methodology of Developing the National Strategic Plan

Policy making of the national strategic plan for rare diseases with virtuosity and consequentiality based on a vision on the existing resources and realistic expectations in the society and according to the priorities in developing macro national strategic plan and after its approval and notification to the Ministries and relevant organizations, developing the operational plan will be conducted by the strategic committee and with the responsibility of Ministry of Health & Medical Education in regard with the strategies and objectives of this national strategic plans. The existing national strategic plan as a macro plan with research standards and utilizing of the precise results of the following proceedings was developed.

- Presenting the processes of developing the national strategic plan, principles and time scheduling
- Establishing expert working groups and conducting expertise studies in different fields
- Analyzing the current status, pathology of the former plans and proceedings, inspecting the weak, strong points, opportunities and threats
- Inspecting the national, comparative and international studies
- Collecting the views and comments of the domestic and international elites and experts, scientific and medical society, authorities and beneficiaries and rare diseases NGOs and communities
- Inspection of the macro strategic and operational national plans, , road maps and reports of rare diseases unions and relevant committees in the world.

Definitions

1- Rare Diseases

A vast group of diseases which have a different prevalence among the countries and varies from 5 to 76 in 100,000 (with a threshold average of 40 in 100,000). Despite the rare diseases impact on a few population, they have a high mental, social and economic burden on the lifestyle of rare patients and their families, they are life threatening and disappointing diseases and need long-term and specialized treatment. About 80% of rare diseases have genetic origin while the rest is non genetic. There are almost 7000 different types of rare diseases and disorders identified in the world. Based on valid definition of the scientific resources in the globe (PubMed), rare diseases are a big group of diseases identified with low prevalence in the population and difficult to diagnose.

2- Genetic Diseases

Genetic diseases are a group of diseases with genetic source, etiology, deficiency, disorder or change in DNA. Genetic diseases may or may not be hereditary (from either of the parents). Genetic diseases could be monogenic, multi-genic or chromosomal.

3- Orphan Drugs

A group of drugs which are mainly used for treatment of rare diseases. Providing the orphan drugs is not cost-effective and due to low demand, the sponsors are not willing to invest on their production plans and projects.

4- Non-communicable Diseases

A vast group of diseases which are not transferred from one person to another and they generally have a long-term and slow treatment process.

5- Congenital Diseases

Congenital diseases are referred to diseases present at birth. They may or may not have genetic or non-genetic origin.

6- Prevalence

It refers to the proportion of population affected with a disease at a particular time period.

7- Incidence

It refers to new cases of a disease which occur in a period of time and in a population at risk.

Policies

Developing a national strategic plan for rare diseases of the country in order to identify and evaluate the specifications of rare diseases by means of a comprehensive method and establishing a political and legal framework including all beneficiaries and the cooperation of the concerned national and regional organizations through forming a national strategic committee supervised by the Ministry of Health & Medical Education and all the beneficiaries as per the following:

- Having access to healthcare services, specifically by taking benefits from the existing healthcare and treatment centers in the country according to the required medical services along with the current plans and making the best use of the capacity of existing structures such as medical clinics, hospitals, healthcare and treatment centers in the form of standard protocols, clustering and classification as well as the referral system for facilitating access to the experts and specialists for diagnosis, treatment and appropriate care including rehabilitation and empowerment the services and also defining healthcare trending for people living with rare diseases.
- Reinforcement the social supports of people suffering from rare diseases by merging the social services with current plans of the Ministry of Cooperatives, Labor & Social Welfare and establishing a referral system among the centers offering healthcare, treatment and social services.
- Supporting national innovative researches
- Collaboration in international research projects
- Identifying, developing and supporting rare patients registry

The main goal of developing the national strategic plan for rare diseases of Iran, is the diagnosis of rare diseases as a national public healthcare priority and defining national intensive care process which will eventually reduce the burden of such diseases on the rare patients, families, caregivers and the healthcare system. The strategy and policy include the national proceedings for precise prevention activities, care enhancement quality at each level of the rare disease, from diagnosis to access and continuity in treatment through the long-term follow up, social care and other services like promoting the innovative rare diseases researches and there should be a defined budget for each required action, accordingly. According to the rarity, distinction and complexity for each of 7000 types of rare diseases, no single country could solve the relevant challenges with rare diseases itself. Hence the pioneering developed countries such as the European countries have an added values to share knowledge and expertise with each other.

Fundamental Principles of The National Strategic Plan for Rare Diseases of Iran

- Collaboration and support by the government
- Defining, coding and identifying all types of rare diseases and registering the rare patients' data
- Research (diagnosis, development of orphan drugs, innovative treatments and the healthcare economy and provision approaches for the orphan drugs
- Establishing the expertise and reference networks
- Development of the social programs and services compatible with the rare diseases
- Empowering the rare diseases NGOs & communities
- Sustainability of the national strategy and guarantee the commitments by the government in the form of 5-year development programs
- Legal, governmental and macro supports

Vision

The vision for developing the National Strategic Plan for Rare Diseases of Iran has upcoming horizon as per following issues:

- Achieving a comprehensive and integrated approach in healthcare and welfare, emphasizing on the human rights and the social justice for rare patients' inclusion
- Determining the rare patients together with other patients as well as paying a special attention to solving their healthcare and treatment needs
- Developing the science, knowledge, technology and promoting healthcare indexes
- Utilizing the capabilities and existing capacities in the country for the patients suffering from rare diseases.

Fundamental Values

Considering the fundamental values & virtues of the Sacred Regime of the Islamic Republic of Iran and with emphasis on Monotheistic Ideology of Islam, ethics, justice and human dignity, the fundamental values of this national strategic plan are as below:

- Deep-rooted belief of the Regime of the Islamic Republic of Iran in providing social justice and health for all the people
- Providing fair and just access to healthcare services as a collective right referring to the principle 29 of the government constitution
- Providing the fundamental needs referring to the principle 43 of the government constitution
- Justice accomplishment in healthcare referring to principles 84 to 94 of the fourth 5-year program Act

Macro Policies

- Applying the capacities and potentialities of the country for reducing the difficulties, troubles and tangles, mortality rate of the rare patients of the country based on the priorities considerations by the Ministry of Health & Medical Education and the available facilities
- Establishing the utmost collaboration with NGOs and patient centered associations in order to make decisions on reducing the difficulties of rare patients, fundraising and financial resources as well as performing executive proceedings to achieve the goals of the national strategic plan for rare diseases of Iran
- Commitment to corporate the social and public responsibility as of the government sovereignty for the rare patients
- Considering the social and public scientific capacity of the country such as the specialists expertise in the related fields of study, educational and research centers, research networks, specialist-science laboratories and active scientific associations in the country for conducting researches and designing national projects according to the relevant priorities and budgets for the rare diseases and discovering new findings as well as achieving evidence-based knowledge
- Establishing inter-sector coordination among all the organizations and departments of the country concerned to consider and optimize suffering

issues with the rare patients such as healthcare, welfare, lifestyle, social and cultural challenges and etc.

- Assigning and developing the principles as well as the international communication frameworks to exchange knowledge and necessary information in order to achieve the efficient approaches for the rare patients upon the existing rules and regulations and observing the principles of the medical ethics
- Continuous and active presence of the beneficiaries at the Ministry of Health & Medical Education and other organizations for drawing the scheme of rare diseases in the country and conducting more accurate pathology of the past proceedings and plans, transparency of the rare patients status through considering the existing structures and plans
- Defining and assigning duties to the responsible rare patients communities and organizations, accordingly
- Assigning the priorities and classifying the services based on the age groups and service packages while identifying the rare diseases in the country and pointing out support limitations for services by the government, insurance companies and NGOs
- Promoting healthcare indexes and social welfare for the rare patients and their families
- Prioritizing prevention proceedings to enhance the status of prevalence and incidence rate of rare diseases based on the rare patients' needs especially at their age of marriage and childbearing
- Applying the crosscut and early returns proceedings to enhance cares based on the rare patients' needs
- Establishing and developing the sustainable policies upon the appropriate legislations, necessary budgeting and certain share allocation out of Gross National Product (GNP)
- Raising awareness of the policymakers, medical society and healthcare experts as well as the public society about the rare diseases

Macro Objectives

- Reducing the risk of incidence and preventing the birth of new cases with rare diseases
- Precise diagnosis of rare diseases based on the prevention principles
- Reducing difficulties, burdens, side effects and mortality due to rare diseases
- Promoting welfare, cultural, social, economic and healthcare conditions for the rare patients and their families through identifying the current resources for a better guide to support with offering special services
- Assuring the rare patient regarding their social justice as well as their benefits from the existing adopted rights
- Providing the education facilities for the rare patients through social supporting, training, programming and empowering the access to higher educational system as well

Macro Strategies

- Developing epidemiologic knowledge about the rare diseases
- Educating and informing the rare patients and their families, healthcare workers, experts and the public society
- Developing rare diseases research by respecting the principles of medical ethics
- Educating healthcare staffs and workers for a better diagnosis of the rare diseases
- Planning for education and prevention from the incidence of new cases in different classes of the society
- Revising the educational and scientific syllabus in the relevant academic levels and inserting new chapter regarding the rare diseases based on the priorities
- Promoting the national and international collaborations for rare diseases by observing the existing rules and regulations
- Empowering the communication networks among the rare diseases
- Developing an organized screening plan with having access to diagnostic laboratories according to the screening factors depending on the cost, efficiency and the policies by the relevant committees
- Promoting the access to treatment and better quality of care services for the rare patients based on the priorities, costs, efficiency as well as focusing on the whole aspects within the research, supplying the orphan

drugs upon the priorities and policies by the Ministry of Health & Medical Education

- Considering the solutions to solve and improve the nutritional deficiencies for some rare patients in need of particular diet by providing better food productions and supplements, establishing the food bank as well as adopting financial supports with a focus on the priorities and policies of the Ministry of Health & Medical Education, accordingly
- Promoting the psychosocial cares for the rare patients and their families
- Utilizing the various aspects like art, health and other related fields for rehabilitating and empowering the mental health, sociocultural characteristics of the rare patients
- Developing and strengthening rehabilitation services in the selected centers to meet the specific needs of the rare patients, providing special facilities as well as considering the social working services
- Implementing through collaboration with the active and dynamic Islamic jurisprudence regarding the termination of pregnancy for fetuses anomalies which are also life threatening with serious birth defects
- Promoting, enhancing and providing the appropriate means of utmost collaboration of NGOs, donators and the culture of social responsibility development of organizations, economic agencies and public societies
- Calling for the attention of all the beneficiaries and rare diseases authorities at public, private sectors and NGOs
- Applying media capacity for attracting public, donors, and policy makers to collaborate
- Raising necessary funds to cover the costs of activities and programing
- Providing the required resources to establish the subspecialty clinical and genetic counselling centers in each province
- Determining scope of duties for the beneficiaries, cooperation and coordination terms and conditions for partners and consensus of opinions among the beneficiaries,
- Forming the strategic board committee of rare diseases at the Ministry of Health & Medical Sciences with the collaboration of the concerned organizations as the Ministry of Cooperatives , Labor & Social Welfare, Ministry of Science, Research & Technology, Planning & Budget Organization, Imam Khomeini (God have mercy upon him) Relief Foundation, Social security Organization, Health Insurance Organization, Islamic Republic of Iran Broadcasting (IRIB) and the elites in various fields as the clinical, basic and social sciences.

Proceedings

- Updating rare diseases registry system (SABNA) to be connected to other electronic files at the rare patients centers such as academic and treatment centers, especially deputy of health office at the Ministry of Health & Medical Education, State Welfare Organization of Iran for monitoring and evaluating disabilities caused by rare diseases and disorders and also avoiding parallel services as well as exchanging data with health insurance electronic systems
- Developing and notifying reports and rare diseases registry programs to signify the healthcare treatment center all over the country
- Coding classification and listing contents for all types of rare diseases in the country,
- Setting up the healthcare system for rare diseases of the country and merging the executive programs with the national strategic plan for rare diseases of Iran through the healthcare network
- Educating and training the personnel for further capabilities and specialties about the rare diseases
- Establishing, operating or utilizing specialized healthcare, treatment and rehabilitation centers in the country for offering healthcare services for the rare patients in case of subject, cost priorities with appropriate time scheduling based on macro policies of MOH-Iran
- Supporting active rare diseases NGOs and associations
- Optimizing mechanisms by making the best of opportunities for a serious and sustainable intervention in offering services to the rare patients and rare diseases researches following the rules and regulations, observing the principles of medical ethics and also identifying patient-centered groups and communities as the main partners
- Raising public awareness and rare diseases service providers regarding their different impacts on the rare patients' lifestyle such as education, family, social communications, ability to work, etc.
- Offering pharmaceutical education to the rare patients and precise raising awareness by the scientific communities, NGOs, rare patient-centered associations, and preventing from the worries and stresses resulted from consuming drugs due to the false advertisement
- Building up connection and effective interaction between the healthcare system of the country and other rare patients service providers
- Providing requirements for the rare patients to collaborate with research projects and other rare diseases activities according to the rules and regulations by observing principles of the medical ethics

- Conducting emergency tests assurance for at high risk individuals when the proper tests are available
- Conducting diagnosis tests in a way that emergency ones to be done at the earliest
- Providing the basic treatment insurance coverage for the rare patients and also supplementary insurance based on the priority of rare diseases and existing funds and credits as well as the certain budgetary and supplying financial resources for drugs insurance coverage and new services in addition to previous excess commitments of the insurance organizations
- Establishment drug bank and specialty pharmacy for rare patients,
- Establishing or exploring the existing rare patients pharmaceutical research centers
- Adopting the relevant Labor laws with labor and supporting employment and its sustainability for the rare patients and their families and customizing workplace for rare patients, accordingly
- Building special parks and welfare, entertainment and cultural centers
- Merging the identification plans for the rare patients based on the application of the healthcare system programs
- Defining the referral processes for the rare patients
- Defining, forming and selecting diagnostic steps: 2nd & 3rd Step
- Defining, forming and selecting treatment steps : 3rd Step
- Merging genetic counselling and prevention plans along with the permanent genetic care for rare diseases in healthcare system based on the social genetic program at the deputy office of the Ministry of Health & Medical Education accompanied by the two strategies of control and prevention of common diseases and rare genetic diseases
- Pursuing the healthcare procedure for the rare patients
- Developing a content of the existing data (nationwide) for the rare diseases
- Developing supportive plans to empower the rare patients' families and to balance their independence, providing welfare and healthcare facilities as well as the occupational work capacity
- Raising public awareness about the hereditary factors of rare diseases (genetic counselling)
- Holding national and specialized events with the concept of solutions for the rare patients' problems and their unmet needs
- Comparing the national strategic plan for rare diseases of Iran with the healthcare programs and existing operational along with prevention and controlling rare genetic diseases as well as merging them with the healthcare system

- Training public society through developing and executing of the educational genetic self-care programs in the families having a rare genetic disease patient and also educating personnel based on distance continuous trainings to prevent the future generation from rare diseases and disabilities

Key Proceedings

- Forming a national strategic plan committee presiding by the Ministry of Health & Medical Education including beneficiaries, main governmental authorities, private and public sectors in charge of the rare diseases to prioritize proceedings and actions based on the importance of the rare diseases, determining the prevalence and burden of disease itself, the costs, existence of effective treatment, the possibility of conducting precise diagnosis methods and accelerating implementation and operation of the national plan in line with the operational plans including the activities and considered proceedings to be classified as well with which the developing and practicing share by the Ministry of Health & Medical Education and other relevant organizations could be merged in this system
- Assigning the specialized rare diseases working groups, sub-groups of the strategic committee attended by the representatives from the governmental, private sectors and rare diseases NGOs & communities
- Forming supervisory and monitoring committee of the national plan, subordinate strategic board committee with the aim of step-ramp pursuit, monitoring and implementation of program, avoiding performance deviation and also as a reference for the viewpoints feedbacks, recommendations, critics, reports presentation to the concerned authorities and conducting revision and rectification process

Organizations & Entities

Ministry of Health & Medical Education, Ministry of Cooperatives, Labor & Social Welfare, Legal Deputy of Presidency, Ministry of Education, Ministry of Science, Research & Technology, Ministry of Economic Affairs & Finance, Ministry of Foreign Affairs, Ministry of Agricultural Jihad, Ministry of Culture & Islamic Guidance, Plan & Budget Organization of Iran, Islamic Consultative Assembly, Social Security Organization, Health Insurance Organization, State Welfare Organization, Islamic Republic of Iran Broadcasting (IRIB), Municipalities, Central Bank of the Islamic Republic of Iran; Imam Khomeini **(God have mercy upon him)** Relief Foundation, Armed Forces (Compulsory Military Service), Red Crescent Society, Rare Diseases Communities, Associations and NGOs, Private & Governmental Universities, Academic, Research, Treatment, Pharmaceuticals and Diagnosis Centers, Pharmaceutical Companies and Scientific Associations

In Memory of The Late Dr. Ali DAVOUDIAN

"Our sincere appreciation to the immense efforts and generous supports by the Late Dr. Ali DAVOUDIAN, the founder of "Rare Diseases Foundation of Iran" (RADOIR). He dedicated his life to establish and develop the National Strategic Plan for Rare Diseases of Iran with generosity, compassion and seriousness."

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- Society for the Protection of PKU Patients
- Society for the Protection of SMA Patients
- Society for the Protection of Cystinosis Patients
- Kootah Ghamatan Boland Hemmat Association
- Neurofibromatosis Association of Iran
- FDA-Iran & MOH-Iran
- Medical Committee of the Rare Diseases Foundation of Iran (RADOIR)
- Rett Syndrome Patients Group
- Gaucher Disease Patients Group
- Myasthenia Gravis Patients Group
- Society for the Protection of MPS Patients
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