IFMSA Policy Proposal
Rare Diseases

Proposed by SfGH U.K and IFMSA- Brazil
Presented to the 69th IFMSA General Assembly August Meeting 2020

SfGH U.K

Name of National Member Organization 1

[Signature]

Sign and stamp of President of National Member Organization 1

IFMSA- Brazil

Name of National Member Organization 2

[Signature]

Sign and stamp of President of National Member Organization 2

Policy Commission
- Agata Oliwa SfGH U.K – 2236127O@student.gla.ac.uk
- Camila Graczyk Corrêa-IFMSA Brazil- vpi@ifmsabrazil.org
- Alaa Abusufian E. Dafallah – ime@ifmsa.org

Draft Policy Proposals have to be sent to all National Member Organizations (nmos@ifmsa.org) by the proposer to request for feedback by June 10 2020 23.59 GMT. Policy Proposals to be discuss at 69th August Meeting General Assembly 2020 have to be sent to gs@ifmsa.org by July 1, 2020 @ 23.59 GMT (please put the code [POLICY] in the beginning of the subject of your email).
Policy Statement

Introduction:

Rare diseases are collectively a common and important global health issue affecting over 350 million people worldwide. In spite of the stark heterogeneity of the conditions, rare disease patients face similar struggles and disadvantages when accessing healthcare including delays in diagnosis, limited access to specialist care and treatment options.

IFMSA position:

Equity in access to healthcare for all is one of the core principles based on which the IFMSA operates. Therefore, we strongly believe that the very nature of the disease a patient lives with should not be an impediment for them to receive high quality care and support.

We are alarmed by the insufficient support for patients living with a rare disease in high income countries (HICs), but even more so in lower- and middle-income countries (LMICs) as well as the relative underrepresentation of teaching on rare diseases within the medical schools curricula. As future healthcare professionals we acknowledge the importance of the awareness of the collective ubiquity of rare diseases and our duty to actively seek opportunities to learn about them throughout our careers. Despite many advances made in the rare disease agenda, we believe that more action at local, national and global level is needed to ensure better quality of care and equal opportunities for patients living with a rare disease. Importantly, we also believe that rare diseases patients are still an especially disenfranchised population in resource-poor settings and action is required to advocate for their needs.

Call to Action:

IFMSA calls for Governments to:

- Establish national multi-disciplinary working groups on rare diseases and develop an evidence-based national policy and strategy for improvement of care for patients with rare diseases, drawing from the experience of other countries (e.g. in the European Union) who have already implemented that successfully
- Start incentivizing the development of medications for rare diseases (“orphan drugs”) through national and international orphan drug policies (or keep reviewing the policy in countries where they already exist)
- Foster inter-country collaborations in negotiations of orphan drug prices with pharmaceutical companies
- Establish international working groups discussing introduction of national and international policies establishing transparent and legally-binding criteria for setting prices of orphan drugs to ensure the prices are not arbitrarily set by pharmaceutical companies
- Support and promote the engagement of patients and patient support groups at every step of the consultation, drafting and implementation of Rare Disease plans and strategies
- Allocate funds to assist the work of patient support groups and finance basic, translational and clinical research and training in rare diseases
- Promote international collaborations between researchers to conduct more robust and meaningful studies on rare diseases
- Support training of international students and researchers from LMICs on rare diseases and genomics, though funded degrees at universities in HICs
- Keep working towards achieving Universal Health Coverage in all countries, keeping rare disease patients in mind
IFMSA calls for NGOs to:
- Continue to support rare disease patients locally, including advice on access to specific patient support groups, social services
- Support international collaborations between patient support groups to share expertise and promote collaborations, especially with organizations with less experience
- Continue to advocate for the rare disease community locally, nationally and globally

IFMSA calls for Medical Schools to:
- Give permission and support students wishing to undertake audits on the quality and quantity of content about rare diseases and genomics in the medical schools’ curriculum
- Establish a realistic curriculum for undergraduate education of medical students on rare disease and genetics based on evidence from audits and experts' opinion, and implement it
- Foster international exchanges of experience and expertise in teaching about rare diseases through conferences and collaborations
- Advertise and facilitate opportunities for medical students to participate in research about rare diseases

IFMSA calls for Global Academic Community to:
- Support medical schools in developing desirable and realistic learning outcomes for undergraduate education of medical students on rare disease and genomics
- Support students in auditing current rare disease and genetics content at their medical school, fostering their interest in research and drive to change what they are passionate about
- Share expertise through international collaborations, e.g. European Reference Networks
- Remember about ethical aspects of research especially genetic research in LMICs and implications of ones' actions for the public's view of the research including possible mistrust
- Ensure diversity of genetic research and foster capacity building for less experienced researchers especially in LMICs, so that they are equipped to take lead in the design, conduct and analyze the data, rather than outsource their local projects to researchers from the HICs

IFMSA calls for Health Professionals and Health Institutions to:
- Continuously seek to keep up to date with new developments in rare diseases especially pertinent to the specialty they work in
- Organize workshops to facilitate further professional development in the area of rare diseases amongst healthcare workers
- Conduct research into social and clinical aspects of rare diseases and offer medical students an opportunity to get involved

IFMSA calls for Health students to:
- Actively seek opportunities to learn about rare diseases, including rare disease patients who are eager share expertise about their condition and the struggles they face
- Set up or support the efforts of the Rare Disease societies at their University to raise awareness about rare diseases including celebrations of the Rare Disease day (29th February)
- Run awareness campaigns about rare diseases and orphan drugs at their University
- Get involved in clinical, scientific and social science research on rare diseases
- Provide constructive feedback on current rare disease teaching events and suggest ideas for improvement to the teaching staff based on own experience

IFMSA calls for the National Member Organizations to:
- Advocate for acknowledgement of the disenfranchised population of patients living with a rare disease with little support especially in LMICs
- Establish an IFMSA working group on Rare Diseases, which aims would be to carry out an audit of rare disease and genomic education in medical schools in its member countries
Position Paper

Background information:

Rare diseases are defined in Europe as those affecting less than 1 in 2,000 people (European Council, 2000), while in the United States it is a disease affecting less than 200,000 individuals countrywide (or 6.25 per 10,000) (Pushpakom et al., 2019). However, since over 7,000 such conditions have been described, this leads to a paradox as although each rare disease is uncommon, together they affect a significant proportion of the population. In fact, it is estimated that collectively around 1 in 17 people in the UK (Muir, 2016) and 1 in 10 people in the US (Kaufmann, Pariser, & Austin, 2018) will be affected by a rare disease at some point in their lives, amounting to over 350 million people around the world living with a rare disease (Shire, 2013).

Even though each rare disease affects patients in a unique way, many struggles they face are strikingly similar. This is not surprising as the reasons behind it stem from the very nature of the diseases, i.e. the small number of patients with each condition and their dispersion around the globe, which makes them prone to not receive sufficient attention. The resulting common challenges include long arduous journey to diagnosis ("diagnostic odyssey"), difficult access to specialist care and limited treatment options (EURORDIS, 2005; Muir, 2016; Shire, 2013).

Overall, patients living with a rare disease face significant barriers in access to and quality of healthcare as well as at an increased emotional and physical disease burden. Therefore, in the context of aspiring to achieve the third Sustainable Development Goal (SDG3) of ‘healthy lives (…) for all at all ages’ (UN, 2019), rare diseases patients, as a disadvantaged population, deserve additional efforts to ensure equitable access to care. Viewing rare diseases as a global health issue, crossing countries’ borders and requiring international efforts, is required in those efforts, especially raising awareness, fostering more robust research efforts and legislature changes, in every country around the globe.

Discussion:

Struggles of patients with (undiagnosed) rare diseases

Rare disease patients face barriers at every step of accessing healthcare and those are best described in European and Northern American patients. In a large survey encompassing the voice of over 12,000 individuals of the former patients, EURORDIS (an NGO uniting the voice of European rare disease patients) investigated their experiences, needs and expectations (EURORDIS, 2009). It confirmed how challenging the pursuit of a diagnosis can be, namely a fourth of surveyed patients had to wait between 5 and 30 years to receive a diagnosis, after having consulted many physicians, underwent many (sometimes unnecessary and invasive) investigations and having suffered from significant psychological and financial burden. During their diagnostic odyssey 40% have received misdiagnoses and some also inappropriate treatments, which not only in itself can be deleterious to health, but also causes further delays in starting appropriate management (if it exists). Even once the correct diagnosis is reached, around a third of the patients are not content with the way it is delivered and how much support was offered. Additionally, despite the fact that 80% of rare diseases have a genetic basis, around 25% of patients were not informed about this and only half received appropriate genetic counselling (EURORDIS, 2009). Therefore, many patients are left to research their disease by themselves and although excellent information and support is available from many patient support groups, during such distressing time it would be reasonable to expect more guidance and support from the clinicians.

As it come to living and managing a rare condition, it is important to know that they are commonly complex and chronic forcing patients to rely on the help from a specialist multidisciplinary team (EURORDIS, 2005). However, again they face multiple challenges with accessing the care they need due the cost and inconvenience of travelling to appointments at distant specialist centers and long
waiting times (EURORDIS, 2009). Combined with the progressive nature of many rare diseases and resulting significant disability, many rare disease patients have limited options for employment. Not uncommonly, both pediatric and adult patients need to rely on their family for support, both financial and in performing daily activities (EURORDIS, 2005).

Effective treatments are available for around 5% of rare disorders (Lancet, 2019), but again their availability varies regionally and nationally. This statistic is particularly striking as around half of rare diseases have an onset in childhood (EURORDIS, 2005) and 30% of these patients die beyond the age of 5 (Lancet, 2019). The dismal prognosis together with isolation and other described challenges all contribute to significant mental health burden for rare disease patients and their families (EURORDIS, 2005). Some parents refuse to give up and become extremely invested in advocating for their children’s condition and liaising with researchers to try to find a cure and in some notable cases these efforts turn into success stories ending with finding new treatments for the condition (e.g. with adrenoleukodystrophy (Lancet, 2008) and alkaptonuria (Findacure, 2019)).

**Additional struggles of patients with undiagnosed rare diseases**

Some rare disease patients might never receive a diagnosis and they will continue to live in anxiety of what future might hold (sometimes called ‘undiagnosed purgatory’). Because of that they endure additional challenges including isolation and helplessness as well as barriers to accessing coordinated medical and social care solely because their problems do not have a name. Additionally, the parents of undiagnosed children face difficult reproductive decisions (EURORDIS, 2009). Such unmet needs of undiagnosed rare disease patients have been recognized in the US by starting the undiagnosed disease programme at the NIH, which is a pioneering initiative attempting to help those patients with finding a diagnosis though thorough evaluations at a national expert center, which are successful in 25-50% of a couple hundred of cases investigated each year (Tifft & Adams, 2014). Other countries like Spain (López-Martín et al., 2018), Italy, Japan, Australia and Canada among others (Taruscio, Floridia, Salvatore, Groft, & Gahl, 2017) have also established similar programmes, however they are still few and far between and their capacity to accept patients is limited.

**Research**

The very nature of being rare means that not uncommonly such conditions do not receive all the research interest and funding they require and if they do many challenges need to be overcome. First and foremost, any research efforts, from describing the natural history or mechanisms of the condition to attempting to devise therapeutic approaches, are hampered by geographical dispersion of patients. Not only does it pose challenges for recruitment, but also for logistics around study appointments and results in many small fragmented studies, which have low statistical power (Kaufmann et al., 2018). Additionally, because of the small population of patients, the competition between pharmaceutical companies and academic institutions for trials participants can be heightened (Ragni et al., 2012).

Therefore, given the scarcity of patients, regional and international collaborations are an essential part of rare disease research (Kaufmann et al., 2018). One organization facilitating such efforts is International Rare Disease Research Consortium set up in 2011 to foster collaborations of multiple funding bodies, industry representatives, academics and patients united by the goal of developing ways to better diagnose and treat rare diseases by optimizing the use of the scarce resources and better coordinating research efforts (IRDiRC, 2013).

Other issues around rare disease research include funding (discussed below) and consideration of patients’ input at all stages. The latter is especially important in assessing the study tolerability, the choice of targeted disease manifestations and final outcomes relevant to patients (Kaufmann et al., 2018). Additionally, attention needs to be devoted to study advertising as around half of 1,200 UK-based rare disease patients recently surveyed felt that they do not get sufficient information about research opportunities into their own condition by their doctors, despite 80% of them being eager to participate. Moreover, only 18% were aware of a registry for their condition, which is another mean of
improving recruitment to clinical trials, therefore increased knowledge of their existence would be desirable (Muir, 2016).

**Orphan drugs**

Development of a new therapeutic takes on average 10 years and costs over 2.5 billion dollars (DiMasi, Grabowski, & Hansen, 2016) and less than 10% of the compounds entering phase 1 clinical trials are eventually approved for marketing and hundreds are discarded at the pre-clinical stages (BIO, Biomedtracker, & Amplicon, 2016). Therefore, investment in any drug development project carries a significant risk for pharmaceutical companies. In case of rare diseases, the risk is compounded by the fact that the target patient population is small and therefore the scope for return of investment and profit for the pharmaceutical companies is limited. Additionally, not only are there thousands of rare conditions and each is caused by a unique pathophysiological mechanism, but also some of the genetic diseases are caused by many different mutations (e.g. Duchenne muscular dystrophy) and thus requiring different targeted therapies. With limited funds, resources and time this only adds to the challenges for development of therapies for rare diseases.

To overcome such barriers, policies incentivizing development of medications for rare diseases (‘orphan drugs’) through offering tax incentives, support with marketing approval and market exclusivity have been introduced. The first, the Orphan Drug Act, was passed in 1983 in the United States (US) (e-CFR, 2019) and similar legislature followed in Singapore in 1991, Japan in 1993 (Kontoghiorghe, Andreou, Constantinou, & Kontoghiorghes, 2014; Pushpakom et al., 2019), in Australia in 1997/8 (Pushpakom et al., 2019) and eventually the Orphan Drug Regulation (No 141/2000) was passed in European Union in 1999 (EuropeanCouncil, 2000). Together with the rapidly developing genomics technologies including affordable and automated genome sequencing and optimized genome editing strategies among others, these policies have allowed for devising new targeted therapies that successfully reached the market. One recent example is onasemnogene abeparvovec (Zolgensma), which is the first gene therapy for spinal muscular atrophy (SMA) for children under the age of two, approved in the US in 2019 (Hoy, 2019) and is being considered by the European Medicines Agency (SMAUK, 2019). Another approach to finding effective therapies for rare diseases is drug repurposing, i.e. finding new uses for already approved medications which are known to be safe. Such approach, therefore, circumvents the need for pre-clinical and early human trials, thus saving both time (taking an estimated 6.5 years) and money (~300 million dollars) (Nosengo, 2016; Pushpakom et al., 2019). In the past, drug reposition relied solely on serendipitous off-target effects, which were further investigated. This approach, although inefficient, resulted in repurposing of, for instance, thalidomide from its initial indication for managing nausea in pregnant women to its effective use in the treatment of two rare diseases, leprosy and multiple myeloma (Pushpakom et al., 2019). Currently, more systematic methods using computational and experimental approaches to drug repurposing exist, which have the potential for bypassing the lacks of knowledge in the pathophysiology of rare diseases, although they have still not delivered the hyped surge in new treatments.

Despite the undeniable potential for drug repositioning to uncover treatments for rare disease, many challenges exist too. They include insufficient sharing of data and compounds, issues with patenting especially if other usable formulation of the drug are already available on the market and limited market exclusivity. Therefore, there is an argument for revising the current drug repurposing legislations in the US and Europe (Pushpakom et al., 2019).

**Funding for available orphan drugs**

As previously described, enormous progress has been in recent years in developing more orphan drugs than ever before. In fact, between 1983 and 2018, the FDA approved over 800 medications for rare diseases (FDA, 2018), for instance alglucosidase alpha for Pompe’s disease and agalsidase beta for Fabry disease. Despite this success, an important challenge has emerged – the drug pricing. Many of the approved medications are very costly with an estimated price of over 200,000$ (Luzzatto et al.,
2018), with the new gene therapy for SMA reaching over 2 million dollars and being the most expensive drug in history (GlobalData, 2019).

Some of the reasons for such a preposterous price are the small market and a need for return of the investment in drug development to the Novartis company. Other reason is the fact that there is no unified worldwide system for assessing the value of a developed drug and thus pharmaceutical companies are free to artificially inflate the prices. Some governments have started requesting release of information about costs incurred for drug development, however without binding legislation, this is only done on a voluntary basis by pharmaceutical companies. Drug prices should be regulated and set based on agreed and legally-binding criteria (Luzzatto et al., 2018). Some of those could include the cost of development, extend of the effect measured by for example added quality-adjusted life years (like used by the UK’s National Centre for Health and Care Excellence, (NICE, 2017)) and the size of the target patient population (Luzzatto et al., 2018). Additionally, where possible counties should collaborate in price negotiations since this significantly increases the market for the medication and therefore the scope for decreasing the price, which has been done successfully in the past by Belgium and the Netherlands (Henrad & Arickx, 2016). Following it would be desirable to include as many countries in such negotiations as possible, e.g. at the level of the European Union, which together comprised a market of over 500 million people (Luzzatto et al., 2018) or also including LMICs too (Cainelli & Vento, 2019).

**Progress in the rare disease agenda in the HIC**

Since the introduction of the notion of ‘rare diseases’ over 40 years ago (Holtzman, 1978), great progress has been made in raising their profile, improving patient care and developing new treatments. Some of the most important milestones that fuelled change were undoubtedly the orphan drug policies. Following on from those, new legislation from the European Union acknowledged the unique needs of rare disease patients and importance of improving their care through a call for introduction of national rare disease policies in the EU member states (EuropeanCouncil, 2009; EURORDIS, 2019). All those achievements were fueled by strong advocacy from determined patient support groups and international NGOs (e.g. EURORDIS, Rare Disease International (RDI)), whose members usually have lived experiences of rare disease and are highly motivated to achieve better access to high-quality care. Thus, progress is largely driven by the patients and their families themselves.

Some of the more recent advances include acknowledgment from the WHO (WHO, 2012) and the recognition of rare diseases as a pressing public and global health issue, both following strong advocacy efforts from patient organizations. Overall, currently rare diseases are a prominent public health priority in most HICs in Europe, North America, Australia and Japan, however, the reality is very different in LMICs.

**Rare diseases in LMIC**

Estimates suggest that among the 6 billion people living in the lower- and middle-income countries (LMICs), over 300 million people are suffering from a rare disease, which is 10 times more than in either Europe or the US (Auvin, Irwin, Abi-Aad, & Battersby, 2018). Resource-poor countries also suffer the highest incidence and burden of congenital anomalies of various etiologies, including rare and genetic causes (Sitkin, Ozgediz, Donkor, & Farmer, 2015; WHO, 2016). Yet, most articles focusing on rare diseases fail to mention anything about the current state of research or management of those conditions in the LMICs. Additionally, a recent review of orphan drug legislations and policies failed to identified any such documents in any Latin American or African country (Gammie, Lu, & Babar, 2015), which adds to the notion that patients living with a rare disease in such under-resourced settings are even more disenfranchised (Cainelli & Vento, 2019). National policies on rare diseases also predominate in the HICs, however other countries around the world are also putting efforts into their development with some examples being Brazil, Argentina, Colombia, Philippines and Fiji (Dharssi, Wong-Rieger, Harold, & Terry, 2017; Shafie et al., 2016).
Further barriers to appropriate care of patients living with a rare disease in the LMICs stem from the fact that most (over 80%) of the genomic data we possess comes from white European and American population (Popejoy & Fullerton, 2016). Limited funding for genomic technologies, staff training and research also contributing to little diversity within the scientific community are partially responsible for this (Bentley, Callier, & Rotimi, 2017). However, it is important not to disregard the another significant barrier, namely mistrust in western researchers due to historical unethical abuses (Bentley et al., 2017; Nature, 2019) and recent disputes over the use of collected patient samples (Eric, 2019; Stokstad, 2019), which can pose a challenge in recruitment for genetic studies in LMICs. In Africa, to overcome some of those barriers, a new collaborative was established called the Human Heredity and Health (H3Africa), which works towards enabling independent genomic research through capacity building and driving new genomics research efforts (Mulder et al., 2018). Additionally and importantly, in December 2019, an important meeting was held in South Africa, the 11th International Conference On Rare Diseases and Orphan Drugs, during which the a call to action was launched for more collaborative work to be undertaken to improve the lives of rare disease patients in the Africa (Baynam et al., 2020).

Still, under current circumstances, introduction of diagnostic exome/genome sequencing for diagnosing rare diseases in LMICs seems like an unachievable goal. In fact, access to even basic and important medicines is limited in many countries, for instance in Tanzania affordability is still an issue for most SCD patients in accessing the disease-modifying hydroxyurea (Luzzatto & Makani, 2019). The goal of improved care for rare disease patients including diagnostic sequencing should, however, not be disregarded as a long term goal, but current focus should be rather put on more feasible measures including nation-wide newborn screening for some rare conditions for which management options are available in Africa (Nkya et al., 2019) to decrease to neonate and child mortality. Another important step would be to establish registries of rare congenital anomalies, given their importance to not only to epidemiological and natural history research of rare diseases, but also to draw more attention to them and facilitate study recruitment in the future.

Overall, in the light of the neglect of rare diseases in the LMICs, the third SDG reading ‘ensuring healthy lives and promoting well-being for all at all ages’ (UN, 2019) is especially pertinent. The goal of making high-quality care available for all patients living with a rare disease is undeniably challenging in resource-poor settings, where changes in the structure of the healthcare system driven by epidemiological and demographic transition force the emphasis to be put on common conditions to ensure efficient use of the scarce available resources (Allotey, Allotey-Reidpath, & Reidpath, 2018). Therefore, in this context, the addition of rare disease to the United Nations’ universal healthcare coverage agenda and declaration adopted by all 193 countries is especially important. Not only does it draw attention to rare diseases at the level of international policy, but also allows the community to demand action from their country’s government and hold it accountable to their declaration (RDI, 2019b).

Devising feasible strategies for improving care of patients with rare diseases while working on achieving universal healthcare coverage for all will be a challenge. That is due to the limited resources available and many other pressing public health issues, some of which might be affecting a larger proportion of the society. Therefore, it is unlikely that rare diseases will be the top of public health agenda, and changes for the patients will not come quickly or easily, however, simply starting to discuss rare diseases and devising some long-term plans is a step forward. International collaboration and exchange of experiences will be essential and some success stories from Colombia, Philippines and Fiji are a useful resource and source of inspiration (RDI, 2019a).

**Teaching and training on rare diseases**

Some of the previously described challenges that rare disease patients face including delays in diagnosis, misdiagnoses and insufficient information about the condition and opportunities to participate in research can be ameliorated by improved education of the (future) healthcare professionals. Until today, no large studies have been conducted to assess how much teaching on rare diseases is included in the undergraduate curricula in medical schools. Published articles on the topic are rather selective with their focus and come almost exclusively from high income countries (HICs), again highlighting the
neglect of the topic in resource-poor settings. Data from Poland suggests that students know very little about rare diseases, e.g. the definition or examples of diseases, although a 30-hour-long facultative course improved their knowledge (Jonas et al., 2017). Non-compulsory courses on rare disease are also offered in other European countries including the UK (Killeen et al. 2016), France (Nourissier, 2010), Ireland (Byrne, 2012) among others (Cismondi et al., 2015) as well as Australia (Elliott & Zurynski, 2015). Many of the courses include input from patients in the teaching sessions, given their expert knowledge of the topic and unique insight (Byrne, 2012; Nourissier, 2010; von Gizycki, 2010). The scarce literature from developing countries not only highlights the underrepresentation of the topic in the curricula, but also underappreciation of the genomics knowledge amongst some healthcare students, which might be driven by limited awareness of its value. This is understandable given more pressing public health issues in resource-poor countries (Muzoriana, Gavi, Nembaware, Dhoro, & Matimba, 2017; Nembaware et al., 2019; Wonkam, Njamnshi, & Angwafo, 2006).

Regarding postgraduate education of the healthcare staff more focus is put on it in HICs, for instance in the UK the National Health Service staff have access to a funded Master’s degree in Genomic Medicine (GEP, 2019). Moreover, some countries emphasize health professional education in their strategies and plans for rare diseases including the UK (NHSEngland, 2018; UKgovernment, 2013, 2019), France (FrenchGovernment 2018), Germany, Brazil and Argentina (Dharssi et al. 2017). The situation is, however, different in many LMICs, which have few or no genetics training programme, limited genetics services and less than 50 genetic counselors in the whole of Africa, most working in South Africa (Abacan et al., 2019). The field is, however, rapidly developing especially in the Asia-Pacific region (Thong, See-Toh, Hassan, & Ali, 2018), South America (Marques-de-Faria, Ferraz, Acosta, & Brunoni, 2004) and in recent years also some African countries (Nembaware et al., 2019) with training programmes and services being developed.

Conclusion

Collectively rare diseases are common, but the scarcity of patients living with individual condition and their widespread geographic distribution pose significant barrier to diagnosis, research and management. Although efforts to tackle those issues are undertaken on national and international level, they are not only nowhere near complete and also mostly centered around Europe and Northern America. Someone once said that ‘families affected by rare diseases represent a medically disenfranchised population that falls through the cracks of every healthcare system in the world’ (Elliott & Zurynski, 2015) and this statement is especially felicitous for rare disease patients living in resource-poor settings.

In the spirit of the third SDG of “ensuring healthy lives (…) for all at all ages”, the inclusion of rare disease in the universal healthcare coverage agenda is a big step in the right direction for improving the care of rare disease patients, no matter where in the world they live in. Access to high-quality healthcare should not be determined by patients’ financial abilities, where they live or what disease they have. In fact, to ensure health equity, special attention needs to be devoted to those who are most disadvantaged and rare disease patients (in developing countries) do belong to this group of patients. While progress towards this goal is going to be challenging and the effects not immediate, given the limited financial, infrastructural and human resources, it is high time that rare diseases receive more attention internationally and are more commonly included in the global health agenda.

References:


Bylaws Paragraphs concerning Policy

Bylaw Paragraph 17.2 Definitions

a. Policy statement: Short and concise document highlighting the position of IFMSA for specific field(s). A policy statement includes neither background information, discussion related to the policy, a bibliography and nor does it quote facts and figures developed by outside sources. The maximum length of a policy statement is 2 pages, including introduction, IFMSA position and call to action.

b. Position paper: A detailed document supporting the related policy statement that contains background information and discussion in order to provide a more complete understanding of the issues involved and the rationale behind the position(s) set forth. A position paper must cite outside sources and include a bibliography.

c. Policy Commission: A policy commission is composed of three people, with 2 representatives of the NMOs and one Liaison Officer. The proposer of the draft is part of the policy commission and is responsible of appointing its members. The tasks of the policy commission are the following:
   i. They are responsible of the quality of the policy document with the approval of the proposer.
   ii. Ensuring the content is based on global evidence.
   iii. Collecting and incorporating NMO feedback after the call for input.
   iv. Coordinating the discussion during the General Assembly.

d. Policy Reviewing Committee: A policy reviewing committee is composed of Vice-President for External Affairs, with 3 representatives of the NMOs. A Policy Reviewing Committee shall submit a report to the Executive Board and the National Member Organizations according to Annex 1. A report shall include the review of all submitted policies and reasons behind the final recommendation.

Bylaw Paragraphs 17.3 -17.7 Adoption of policies

- A draft policy statement, position paper and the composition of the policy commission must be sent to the NMO mailing list by the proposer in accordance with Annex 1. Input from NMOs is to be collected between submission of the draft and submission to the General Secretariat.
- The final policy statement and position paper are to be sent in accordance with paragraph 9.4, using the template provided in the call for proposals. The proposal must be co-submitted by two NMOs from different regions or the Team of Officials. A corrected version of this document may be submitted according to paragraph 9.5. Correction may not be used to add members to the policy commission.
- Policy statements and position papers must be presented to NMOs during the first working day of the IFMSA General Assembly.
- A motion to adopt the policy statements and position papers must be submitted the day before the relevant plenary by two NMOs from different regions or an IFMSA Official, the IFMSA Team of Officials or the IFMSA Executive Board. Adoption requires ⅔ majority.
- Amendments may be sent to the proposer in accordance with Annex 1. Amendments made during a General Assemblies or after the deadline stipulated in Annex 1, shall be submitted to the Chair at the latest 23:59 observed in the timezone of the relevant General Assembly on the day before the scheduled start of the session in which the policy will be voted on. These amendments require ⅔ majority to pass.