Subjects Discussed Briefly During Zoom Conference Sessions

1. Undiagnosed Diseases Network International and Developing Countries Working Group
2. Underserved and Indigenous Populations in LatAm and rare diseases in Chile
3. Indigenous Populations
4. Promoting Implementation of Genomic Medicine in Low Middle Income Countries and Global Genomic Medicine Collaborative (G2MC) Flagship Project Genomic Medicine Clinics for Rare Genetic Disorders and Clinical Genetic Services in Ecuador Opportunities and Needs
5. Rare Diseases Opportunities and Needs in Colombia
6. Rare Diseases Opportunities and Needs in Argentina
7. Regeneron Genetics Center
8. Call for Volunteers for the Steering Committee and the Working Groups

A framework of possible areas of emphasis is provided as a starting point:

- Research Infrastructure – National Legislation, Needs Assessment, and Health Care, Research, and Regulatory Policies
- Research Collaboration/Research Networks and Consortia
- Technology Advances – Telehealth, Telemedicine
- Training of Research Investigators and Clinicians
- Clinical Care and Treatments
- Diagnosis and Genomic Advances
- Underserved populations such as indigenous people who are economically disadvantaged and under-reported.
- The Expanding Role of Patient Advocacy Groups
- Preclinical Innovation, Product Discovery, and Development
- Impact of COVID-19 Pandemic on Rare Diseases Research – Present and Future Concerns and Other Opportunities

We inspire global collaboration among all rare disease stakeholders

Outline of Possible Planned Activities of Enfermedades Raras en el Caribe y América Latina (ERCAL) ICORD Initiative (Other Potential Partners include UDNI, RDI IRDiRC and G2MC)

a. Identify Steering Committee and Issue-Specific Work Groups
b. Utilize Steering Committee to Identify Framework Issues
c. Utilize Work Groups for Specific Issues and Areas of Interest
d. Prepare Draft Report
e. Gain Acceptance of Final Report at a Future ICORD meeting
f. Publication of Final Report

Links to Reports from Previous ICORD Activities

1. A call for global action for rare diseases in Africa
   Baynam GS, Groft S, van der Westhuizen FH, Gassman SD, du Plessis K, Coles EP, Selebatso E, Selebatso M, Gaobinelwe B, Selebatso T, Joel D, Llera VA, Vorster BC, Wuebbels B, Djoudalbaye B, Austin CP, Kumuthini J, Forman J, Kaufmann P, Chipeta J, Gavhed D, Larsson A, Stojiljkovic M, Nordgren A, Roldan EJA, Taruscio D, Wong-Rieger D, Nowak K, Bilkey GA, Easteal S, Bowdin S, Reichardt JKV, Beltran S, Kosaki K, van Karnebeek CDM, Gong M, Shuyang Z, Mehrian-Shai R, Adams DR, Puri RD, Zhang F, Pachter N, Muenke M, Nellaker C, Gahl WA, Cederroth H, Broley S, Schoonen M, Boycott KM, Posada M. Nat Genet. 2020 Jan;52(1):21-26. doi: 10.1038/s41588-019-0552-2. No abstract available.

2. 11th International Conference on Rare Diseases and Orphan Drugs, Cape Town South Africa.

3. The need for worldwide policy and action plans for rare diseases
   Forman J, Taruscio D, Llera VA, Barrera LA, Coté TR, Edfjäll C, Gavhed D, Haffner ME, Nishimura Y, Posada M, Tambuyzer E, Groft SC, Henter JI; International Conference for Rare Diseases and Orphan Drugs (ICORD).
   Acta Paediatr. 2012 Aug;101(8):805-7. doi: 10.1111/j.1651-2227.2012.02705.x. Epub 2012 May 11. PMID: 22519914 Free PMC Article
   Yukiwariso Declaration on the ICORD website

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Framework of Possible Areas of Emphasis – Expanded Discussions

1. Research Infrastructure – National Legislation, Needs Assessment, and Health Care, Research, and Regulatory Policies

    - Consider Needs Assessments of patients, families, patient advocacy groups, health care providers, researchers, industry, payers, government regulatory and research agencies, policy makers, education providers in partner countries are similar
    - Assess current initiatives, clinical trials, patient registries, natural history studies, and information sources to utilize and participate on a global basis
    - Utilize international models to emphasize and develop policies and legislative initiatives, regulations, national and strategic plans, research, regulatory decision-making, health care services and reimbursement procedures to consider in individual countries

2. Research Collaboration

    - Data sharing from treatment and research resources
    - Specialty clinics at dedicated centers of excellence should expand emphasis and provide appropriate research infrastructure for clinical evaluations and hubs of optimal patient-centered care.
    - Ready access to collaborative multi- and interdisciplinary research and treatment teams who communicate with one another is essential.
    - Develop and Promote Research and Product Development infrastructure in the many countries having good science and biopharmaceutical capabilities
    - Sharing of data and bio-specimens, patient engagement activities, and sustainable and scalable research models are essential to translate the advances in genomics, clinical phenotyping, epidemiology, functional studies, environmental research, imaging procedures, bioinformatics, health informatics, and communications technology to deliver better prevention, diagnostics and novel therapeutic interventions.
    - Information from indigenous populations contributes to an increased understanding of the relationship between genetic variations and disease.

3. Technology Advances

- Assess Telemedicine and Telehealth initiatives to link all communities with Tertiary Care Centers
- Advanced genetic and diagnostic techniques should become available and applicable in developing countries to facilitate diagnosis. Information about genetic variants should be made publicly available from different regions of the world.
- Utilize social media or mobile technologies such as smart phones to establish active partnership throughout the research process.

4. Training of Research Investigators and Clinicians

- Provide training programs for Health Care Provider specialists, clinical geneticists, genetic counselors, clinical guidelines, more tertiary centers or centers of expertise
- Clinical Trial Design and Analysis courses with regulatory partnership needs and emphasis
- Training for health care providers in Clinical Medicine, Genomics, Clinical Trial Design and Data Analyses
- Big Data Analytics

5. Clinical Care and Treatments

- World Health Organization's (WHO) and Pan American Health Organization (PAHO) programs to ensure that all people have access to diagnostic, preventive, curative and rehabilitative health services
- Third Sustainable Development Goal (WHO) is to ensure healthy lives and to promote well-being across the lifespan and provide access to appropriate treatment and care facilities
- Identify and expand Referral Pathways from primary care providers to Tertiary Care Centers.
- Comprehensive Newborn Screening Programs are an essential part of public health efforts to address rare diseases.
- International Clearinghouse for Birth Defects Surveillance and Research (ICBDRS) is affiliated with the WHO and has as its mission to join birth defect reporting programs

from around the world with the aim of conducting worldwide surveillance and research to prevent birth defects and to ameliorate their consequences

- Surveillance procedures should aim to achieve much higher reporting rates for Congenital Malformations. Inadequate surveillance has hampered appropriate allocation of resources based on the information generated.
- Meet supply chain demands of irregular supply of medicine due to poor storage conditions, short shelf life, limited size of orders and the cost of international transport
- Expand awareness, advocacy, and outreach to everyone including those with low income, poor literacy, minority ethnic status, and living in underserved and marginalized populations in both urban and rural areas.

6. Diagnosis and Genomic Advances

- Access to an early, timely and correct diagnosis should be an achievable goal for all populations in all nations. Successful diagnostic work is based on multidisciplinary teamwork, consisting of clinical geneticists, different medical specialists along with genetic counselors and bioinformaticians.
- Improve limited Research capacity and health system resources and eliminate researchers working in isolation.
- Information gathered from undiagnosed patients should be shared with organizations with a focus on undiagnosed diseases such as the Undiagnosed Diseases Network International while respecting patients' wishes for privacy.
- Participating in existing or newly created undiagnosed disease programs with access to improved sequencing, and other diagnostic, capabilities should help increase the

potential for a correct diagnosis. All currently un-diagnosable individuals will enter a globally coordinated diagnostic and research pipeline. (IRDiRC)

7. Expanding Role of Patient Advocacy Groups

- Informed patients and families should be accepted as knowledgeable sources of information, especially in Emerging and Low- and Middle- Income Countries (LMIC).
- The need for Professionalization of the patient associations, with training, tools, and templates and support for national organizations to develop local groups
- Patient centricity continues to gain acceptance in the research and community settings.
- Strengthen Patient Advocacy Community with recognition as critical partners in Research and Care
- Countries should encourage global patient advocacy group collaborations, e.g. African Alliance for Rare Diseases, Genetic Alliance South Africa, IRDiRC, ICORD, Rare Diseases International

8. Preclinical Innovation, Product Discovery and Development

- De-risking potential drug targets or research projects
- Make natural products from indigenous locations more attractive for commercial investment.
- High-throughput screening technology
- Well-designed chemical libraries
- Analytical chemistry,
- Assay development,
- Late-stage pre-clinical Development Activities, including:
- Systems biology,
- Chemical synthesis and optimization,
- State-of-the-art informatics capabilities,
- Regulatory requirements for drug development

9. Underserved Populations Such as Indigenous People Who Are Economically Disadvantaged and Are Underreported

- Consider the genomic/ phenotypic data from these groups as very important

- Include Cultural and familial approaches that provide novel solutions e.g., think of family/ communal consent via blockchain methods
- Discuss other aspects of more culturally appropriate care to improve service delivery, etc. These will then provide solutions for everyone, Indigenous or not

10. Impact of COVID-19 Pandemic on Rare Diseases Research – Present and Future Concerns and Opportunities

- Recognise the many opportunities for RD coming out of COVID that can be progressively actioned and capitalised on, especially in the peri-COVID and (health) system recovery phases

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Selected References and Abstracts with Links to Published Articles

1. Review of 11 national policies for rare diseases in the context of key patient needs. Dharssi S, et al. Orphanet J Rare Dis. 2017. PMID: 28359278 Free PMC article. Review.
   Despite these legislative efforts and the growing contributions of patient advocacy groups in moving forward implementation and adoption of rare disease programs, gaps still exist across the policy landscape for several countries. ...Further, we found that while national rare disease plans provide important guidance for improving care, implementation of plans is uneven across countries. ...
   Included in Article Rare Diseases Policies in 11 countries: Germany, France, the United Kingdom, Canada, Bulgaria, Turkey, Argentina, Mexico, Brazil, China, and Taiwan.

2. A compilation of national plans, policies and government actions for rare diseases in 23 countries. Khosla N, et al. Intractable Rare Dis Res. 2018. PMID: 30560012 Free PMC article. Review.
   The purpose of this scoping review is to examine and compare published reports on national plans, polices and legislation related to all rare diseases in different countries. ...Multinational programs supported by common or similar laws are likely to have a greater impact on rare diseases than single country programs....

Table 1. (Included in Article)

Rare disease plans, legislation, programs or strategies in Latin American Countries

Country	Definition of rare disease	National Plan	Legislation	Program or Strategy	Highlights
Argentina (25,38,39)	EU Definition	No	Yes	Yes	*Law 26.689 (2011)*: Intended to help rare disease patients and their caregivers by promoting the development of patient registries and screening programs, and educational and social support activities.
Brazil (25,39,40)	No more than 65 cases per 100,000 people.	No	Yes	Yes	*"National Policy for Rare Diseases"(2014):* Includes equal healthcare services, create care guidelines for these patients at every stage of a Unified Health System care, offers comprehensive care in the Health Care Network, improves universal and regulated access for rare disease patients, ensures access to care, and quality healthcare.
Chile (39)	EU Definition	No	Yes	N/A	The Ricarte Soto law aims at providing funding for care of rare disease patients. It assigned a grant of 200 billion pesos for 4 years.
Colombia (39,41,72)	1 case per 5,000 people.	No	Yes	N/A	*Law 1392 (2010)*: Identifies the lack of orphan drugs as a health issue impacting the healthcare system. Considers social protection policies. It contemplates creating a registry for rare disease patients and collaborating globally for research.

Country	Definition of rare disease	National Plan	Legislation	Program or Strategy	Highlights
Mexico (25,39)	EU Definition	No	Yes	No	*Article 224 revision (2012)*: Recognizes orphan drugs and their treatments. Ministry of Health may enforce market authorization, no rules for market exclusivity.
Peru (38,39)	No Definition	No	Yes	N/A	*Law 29698 (2011)*: Includes diagnosis, surveillance, prevention, care and rehabilitation.

3. Access to Orphan Drugs: A Comprehensive Review of Legislations, Regulations and Policies in 35 Countries. Gammie T, et al. PLoS One. 015. PMID: 26451948 Free PMC article. Review.
   The majority of countries (27/35) had in place orphan drug legislation. Access to orphan drugs depends on individual country's pricing and reimbursement policies, which varied widely between countries. ...Importantly, China and India, two of the largest countries by population size, both lack national legislation for orphan medicines and rare diseases, which could have substantial negative impacts on their patient populations with rare diseases....

4. Improved Diagnosis and Care for Rare Diseases through Implementation of Precision Public Health Framework. Baynam G, et al. Adv Exp Med Biol. 2017. PMID: 29214566 Review.
   Consequently we are witnessing a paradigm shift in public health policy and practice towards 'precision public health'.Patient and stakeholder engagement has informed the need for a national public health policy framework for rare diseases. ...Knowledge sharing is critical for public health policy development and improving the lives of people living with rare diseases....

5. Key outcomes from stakeholder workshops at a symposium to inform the development of an Australian national plan for rare diseases. Molster C, et al. Orphanet J Rare Dis. 2012. PMID: 22883422 Free PMC article.
   At present, Australia does not have a national plan for rare diseases. To progress such a plan an inaugural Australian Rare Diseases Symposium was held in Western Australia in April 2011. ...The translation of the symposium outcomes to government policy (i.e. a national plan) requires the consideration of several factors such as the under-

representation of some stakeholder groups (e.g. clinicians) and the current lack of evidence required to translate some of the symposium outcomes to policy options. ...

6. Bridging the Gap between Health and Social Care for Rare Diseases: Key Issues and Innovative Solutions. Castro R, et al. Adv Exp Med Biol. 2017. PMID: 29214594 Review.
   Bridging the gaps between health and social care for rare diseases is not only necessary but crucial to increase the life expectancy, quality of life and autonomy of people living with a rare disease, supporting them in the full realisation of their fundamental human rights. The complexity of rare diseases, their strong relation to disability and the current unmet social and daily life needs of people living with a rare disease must not be underestimated and require urgent attention from all stakeholders involved in care provision, from healthcare to social and community services. The Commission Expert Group Recommendations to Support the Incorporation of Rare Diseases into Social Services and Policies, adopted unanimously in April 2016, by the representatives of European Member States and the other rare disease stakeholders, clearly set the tone for the need to promote measures that facilitate multidisciplinary, holistic, continuous, person-centred and participative care provision to people living with rare diseases. These recommendations, sided by other recent policy developments at European and national levels, represent an important policy step into approaching rare diseases' complex challenges in regards to holistic care provision. Innovative approaches aiming at bridging the gap between health, social and community service and support providers are currently being developed and tested in different European countries: standards of care, networks of expertise, case management services, one-stop-shop services, amongst others. These ongoing pilot approaches, presented in this chapter, have the power to inspire future policies and the effective and efficient implementation of holistic care pathways for people living with a rare disease, bringing about significant changes for patients, carers, care providers, competent authorities and the society at large. Nonetheless, the challenges to fully address this issue remain numerous and other key issues will also need to be taken into account when moving forward with the implementation of measures that aim at bridging the gaps between care providers and providing holistic care to people living with a rare disease....

7. Rare disease policies to improve care for patients in Europe.
   Rodwell C, et al. Biochim Biophys Acta. 2015. PMID: 25725454 Free article.
   These initiatives contributed to the development of rare diseases policies at European and national level aimed at improving care for patients with rare diseases. A review of the political framework at European level and in European countries is provided to demonstrate how legislation has created a dynamic that is progressively improving care for patients with rare diseases. ...

8. Comparative Analysis of Legislative Requirements About Patients' Access to Biotechnological Drugs for Rare Diseases in Central and Eastern European Countries.
   Kamusheva M, et al. Front Pharmacol. 2018. PMID: 30079023 Free PMC article.
   Objectives: The aim of the study was to compare the access of patients with rare diseases (RDs) to biotechnological drugs in several Central and Eastern European countries (CEECs). ...The access to BOMPs is similar among the observed CEECs and the countries with the best access are Hungary and Greece. The influence of BOMP expenditures on the budget in the individual countries is significant....

9. Tackling rare diseases at European level: why do we need a harmonized framework?
   Taruscio D, et al. Folia Med (Plovdiv). 2007. PMID: 18018471
   So far only France has developed a national strategic plan for rare diseases, Bulgaria is in the process of approving its national plan for RD and Spain is in the process of developing it. ...The elaboration of a European Commission Communication on rare diseases will ensure that common policy guidelines are shared everywhere in Europe. ...

10. Challenges raised by cross-border testing of rare diseases in the European union.
    Pohjola P, et al. Eur J Hum Genet. 2016. PMID: 27381091 Free PMC article.
    As the availability of genetic tests has grown rapidly during the last decade along with the increasing knowledge of the genetic background of rare inherited diseases, sending DNA samples to another country for analysis has become more of a routine than an exception in clinical diagnostics. ...In addition, the differences between countries regarding the reimbursement and authorization policies of cross-border testing were significant, thus confirming the pre-existing assumption about unequal access to genetic testing in the different Member States. ...

11. A cross-national comparative study of orphan drug policies in the United States, the European Union, and Japan: towards a made-in-China orphan drug policy. Liu BC, et al. J Public Health Policy. 2010. PMID: 21119648
    In several countries--the United States, the EU, and Japan--specific legislation has been enacted to encourage pharmaceutical companies to expedite the development of drugs for rare diseases, orphan drugs, and to assure access to them. We analyze the strengths and weaknesses of the incentives in these laws and describe the status of rare diseases in China. We offer some recommendations for orphan drug legislation in China, based on local research on rare diseases....

12. [Global strategy for rare and intractable diseases] Kawashima Kodama T. Rinsho Shinkeigaku. 2013. PMID: 24291960 Japanese.
    Along with legislation or regulation of orphan drugs development, treatment and care for rare diseases have been emphasized in each national healthcare system globally. In the US, the Office of Rare Diseases was established under NIH in 1989 and European countries also started collaboration for rare disease projects with their

13. A comparative study of European rare disease and orphan drug markets.
Denis A, et al. Health Policy. 2010. PMID: 20800761 Review.
RESULTS: These countries adopted varying approaches towards regulating rare disease and orphan drug markets and, hence, the availability, pricing and reimbursement of orphan drugs vary between countries. ...CONCLUSIONS: Extensive government intervention exists in rare disease and orphan drug markets in the countries studied. Our recommendations are to define priorities for research on rare diseases and orphan drugs at the European level, to set up disease and patient registries with a view to investigating the long-term effectiveness and cost-effectiveness of orphan drugs, to assess the profitability of orphan drugs, and to take into account societal considerations when evaluating orphan drugs....

14. Principles for consistent value assessment and sustainable funding of orphan drugs in Europe. Gutierrez L, et al. Orphanet J Rare Dis. 2015. PMID: 25935555 Free PMC article.
The European Orphan Medicinal Products (OMP) Regulation has successfully encouraged research to develop treatments for rare diseases resulting in the authorisation of new OMPs in Europe. ...Against this background, policy makers in many countries are considering reforms to improve access to OMPs. This paper proposes ten principles to be considered when undertaking such reforms, from the perspective of an OMP manufacturer. ...

15. Ethics, genetics and public policies in Uruguay: newborn and infant screening as a paradigm. Larrandaburu M, et al. J Community Genet. 2015. PMID: 26021874 Free PMC article.
Uruguay is a middle-income country and the smallest in South America. Its population is under 3.3 million. The demographic and epidemiological characteristics are similar to those of developed countries, with a high burden associated with congenital anomalies. ...The Ministry of Public Health created the Comprehensive Plan on Birth Defects and Rare Diseases (PIDCER), to develop a strategic public policy tool enabling comprehensive, universal, quality care during their entire lifetime. ...

16. A civil society view of rare disease public policy in six Latin American countries.
Mayrides M, et al. Orphanet J Rare Dis. 2020. PMID: 32106873 Free PMC article. Review.
Several recently published reviews highlight the importance of country efforts to address rare diseases and orphan drugs policy comprehensively. ...This allowed us to identify five broad policy categories for subsequent analysis: national laws, national regulations, health system incorporation of rare disease treatments, care delivery, and patient engagement. By describing the different approaches, challenges and timelines across six countries, our research demonstrates that strengthening rare disease policy first requires a common understanding and local consensus of each country's recent past and current situation. ...

17. Orphan drug development across Europe: bottlenecks and opportunities.
Heemstra HE, et al. Drug Discov Today. 2008. PMID: 18583178 Review.
With the assignment of the 500th European Union orphan drug designation in 2007, the Regulation on Orphan Medicinal Products truly begins to show its potential for delivering new medicines to patients with rare diseases. ...Our findings underline the importance of innovation-based policies to enhance the development of orphan drugs in Europe....

18. Achieving optimal cancer outcomes in East Africa through multidisciplinary partnership: a case study of the Kenyan National Retinoblastoma Strategy group.
Hill JA, et al. Global Health. 2016. PMID: 27229322 Free PMC article.
Take for example the aggressive early childhood eye cancer retinoblastoma, where survival reaches 97 % in resource-rich countries, but is as low 30 % in some resource-limited nations, where 92 % of the burden lies. ...Elements of the Kenyan National Retinoblastoma Strategy may be useful to other developing countries struggling with limited survival of retinoblastoma and other cancers or rare diseases....

19. International perspectives on newborn screening. Pollitt RJ. J Inherit Metab Dis. 2006. PMID: 16763907
Different countries are introducing the technology at different rates and for different disease panels. Current policies in the United Kingdom, Germany and the United States are taken as examples. ...The lack of even broad concordance at the level of national policy is extremely disturbing. Though all discussion is nominally founded on the ten principles laid down by Wilson and Jungner in 1968, there seems no generally accepted way of using these principles, or derived criteria, as objective decision tools. ...

20. Pharmaceutical lobbying in Brazil: a missing topic in the public health research agenda. Paumgartten FJ. Rev Saude Publica. 2016. PMID: 28099661 Free PMC article.
In the US, where registration of lobbyists is mandatory, the pharmaceutical industry and private health-care providers spend huge amounts of money seeking to influence health policies and government decisions. ...The main goals of Interfarma lobbying activities are: shortening the average time taken by the Brazilian regulatory agency (ANVISA) to approve marketing authorization for a new drug; making the criteria for incorporation of new drugs into SUS (Brazilian Unified Health System) more flexible and speeding up technology incorporation; changing the Country's ethical clearance system and the ethical requirements for clinical trials to meet the need of the innovative drug industry, and establishing a National Policy for Rare Diseases that allows a prompt incorporation of orphan drugs into SUS. ...

21. Challenges to orphan drugs access in Eastern Europe: the case of Bulgaria.
Iskrov G, et al. Health Policy. 2012. PMID: 22939047
This article explores how an Eastern European country could deal with orphan drugs access, combining EU policies with its own national settings. ...The analysis identifies four

important challenges to orphan drugs' access in Eastern Europe: (1) elaboration of new orphan drugs pricing approaches, (2) further interaction of cost-effectiveness analysis with medical criteria, (3) active introduction of epidemiological registries for rare diseases, and (4) research of societal preferences and raising public awareness....

22. International Charter of principles for sharing bio-specimens and data.
Mascalzoni D, et al. Eur J Hum Genet. 2015. PMID: 25248399 Free PMC article.
This is especially evident for rare disease research. Currently, the rising value of data and bio-specimen collections does not correspond with an equal increase in data/sample-sharing and data/sample access. Contradictory legal and ethical frameworks across national borders are obstacles to effective sharing: more specifically, the absence of an integrated model proves to be a major logistical obstruction. ...

23. Occupational health and safety in the least developed countries--a simple case of neglect. Ahasan MR, et al. J Epidemiol. 2001. PMID: 11388496 Free article.
Steps to control work exposure limits have been ineffective, since national policies have been rare, owing to the multiple obstacles in preventing occupational problems. ...Examples are likewise given to show the real situation of health and safety in the least developed countries....

24. A cross-national comparison of orphan drug policies: implications for the U.S. Orphan Drug Act. Thamer M, et al. J Health Polit Policy Law. 1998. PMID: 9565894
Six countries--Canada, France, Japan, Sweden, the United Kingdom, and the United States--were studied to compare public policies affecting the development and marketing of pharmaceuticals for rare diseases (i.e., orphan drugs). Information was obtained from a variety of published and unpublished sources, including interviews with public policy and pharmaceutical experts in each country. ...