The International Rare Diseases Research Consortium (IRDiRC) has created a quality label, ‘IRDiRC Recognized Resources’, formerly known as ‘IRDiRC Recommended’. It is a peer-reviewed quality indicator process established based on the IRDiRC Policies and Guidelines to designate resources (ie, standards, guidelines, tools, and platforms) designed to accelerate the pace of discoveries and translation into clinical applications for the rare disease (RD) research community. In its first year of implementation, 13 resources successfully applied for this designation, each focused on key areas essential to IRDiRC objectives and to the field of RD research more broadly. These included data sharing for discovery, knowledge organisation and ontologies, networking patient registries, and therapeutic development. ‘IRDiRC Recognized Resources’ is a mechanism aimed to provide community-approved contributions to RD research higher visibility, and encourage researchers to adopt recognised standards, guidelines, tools, and platforms that facilitate research advances guided by the principles of interoperability and sharing. This paper presents an overview of the resources that have obtained the ‘IRDiRC Recognized Resources’ designation after the first year of its implementation.

**IRDiRC Recognized Resources**

The application for ‘IRDiRC Recognized Resources’ is open to all project leaders of different resources (ie, standards, guidelines, tools, and platforms) designed to advance RD research and development. Applications are peer-reviewed with rolling submission based on a number of criteria (Table 1), and in particular, two mandatory requirements: resources must be within IRDiRC’s focus and mission as defined in the IRDiRC Policies and Guidelines document, and have multinational connectivity and audience. Eligible resources include software, bioinformatics platforms, web services, RD data or biospecimen collections, international standards, and international guidelines (Table 2). Other resources, although acknowledged as important, are not eligible for the recognition: national, regional, or institutional biobanks and registries for RD; resources that are dedicated to a single disease entity; resources that could provide some utility for RD, but are primarily designed for broader use; and commercial resources.

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Received 31 August 2016; accepted 6 September 2016; published online 26 October 2016
In its first year, 13 resources have been given the ‘IRDiRC Recognized Resources’ designation. These resources include three guidelines, five platforms, two reference databases, two standards, and an advisory committee (Table 3). These resources are hereby presented according to the area of focus in the context of the goals and objectives of IRDiRC.

**Facilitating international data sharing for discovery**

Four recognised resources have a strong emphasis on sharing approaches, aiming to advance knowledge about RD by encouraging global scientific collaboration while respecting ethical considerations. Two of these resources set out guidelines for legally- and ethically-grounded sharing. The ‘International Charter of principles for sharing bio-specimens and data’ and the ‘Framework for responsible sharing of genomic and health-related data’ aim to remove bottlenecks for effective sharing of RD data without compromising privacy, consent, and interest of individuals who participate and contribute their data for biomedical research. PhenomeCentral is a repository of RD patient phenotypic and genetic data, deposited by clinicians and scientists in this secure database to facilitate discovery; its collaborative model aims to understand the clinical spectrum and underlying mechanism of RD. In a similar manner, DECIPHER is a database that enables the depositing, analysing, and sharing of phenotype-linked plausible variation in patients with RD, thereby empowering diagnosis and discovery. Both PhenomeCentral and DECIPHER are participants of the Matchmaker Exchange initiative, a collaboration between IRDiRC and the Global Alliance for Genomics and Health to enable genomic discovery through the exchange of phenotypic and genotypic profiles using a federated platform, thereby supporting the IRDiRC goal of providing a means to diagnose most RDs by 2020.

**Knowledge organisation and ontologies**

Five widely used recognised resources are found in the area of knowledge organisation and ontologies. With respect to knowledge organisation, two databases facilitate access to RD information, Orphanet, and the Online Mendelian Inheritance in Man (OMIM). Orphanet is a reference portal for RD and orphan drugs. It provides comprehensive information on research projects and their funders, registries and biobanks, platforms for research, clinical trials, a nomenclature and classifications of RDs, genes, and associated phenotypic and epidemiological data. OMIM is a widely used knowledge base of human genes and associated phenotypes comprised of over 23 000 structured free-text entries.

**Table 1 Assessment criteria for ‘IRDiRC Recognized Resources’**

| Mandatory criteria | Additional criteria |
|--------------------|---------------------|
| Within IRDiRC’s focus and mission | Functional and accessible with minimal downtime |
| Multinational connectivity and audience | Development and maintenance team |
|                       | Clear and well-documented terms of use and license policies |
|                       | Adheres to all relevant ethical and privacy policies and requirements |
|                       | Process in place for quality control and life cycle management |
|                       | Undergoes scientific peer review |
|                       | Financially viable for the following 3 years |
|                       | Documents its core impacts (eg, number of users, number of visits, etc) |
|                       | Demonstrates relevant and ongoing activity in sharing and dissemination |

**Table 2 Eligibility for different types of ‘IRDiRC Recognized Resources’**

| Eligible resources | Excluded resources |
|--------------------|--------------------|
| Software, bioinformatics platforms, and web services | National, regional, or institutional biobanks RD, or a single disease entity |
| Data collections/biospecimen collections | National, regional, or institutional registries for RD, or a single disease entity |
| International standards | Resources with some utility for RD research, but primarily designed for broader use |
| International guidelines | Commercial resources |

**Orphanet**

Orphanet is a reference portal for RD and orphan drugs. It provides comprehensive information on research projects and their funders, registries and biobanks, platforms for research, clinical trials, a nomenclature and classifications of RDs, genes, and associated phenotypic and epidemiological data. OMIM is a widely used knowledge base of human genes and associated phenotypes comprised of over 23 000 structured free-text entries.

**European Journal of Human Genetics**
Facilitating international data sharing
International Charter of principles for sharing bio-specimens and data Guideline The Charter provides recommendations for successful legally- and ethically-grounded sharing of bio-specimens and data
Framework for responsible sharing of genomic and health-related data Guideline The Framework provides a principled and practical framework for the responsible sharing of genomic and health-related data
PhenomeCentral Platform PhenomeCentral is a repository for secure sharing of phenotypic and genotypic data in the RD community, thereby connecting patient profiles
DECIPHER Platform DECIPHER is a database and web-based platform enabling the deposition, analysis, and sharing of phenotype-linked plausibly pathogenic variation in patients with RD

Knowledge organisation and ontologies
Orphanet Reference/database Orphanet is a reference portal for information on RD and orphan drugs
Online Mendelian inheritance in man (OMIM) Reference/database OMIM is a database of human genes and genetic phenotypes comprised of over 23 000 structured free-text entries
Orphanet rare disease ontology (ORDO) Platform ORDO provides a structured vocabulary for RD, thereby aiming to define relationships between diseases, genes, and other features of interest
Human phenotype ontology (HPO) Standard HPO provides a standardised vocabulary of phenotypic abnormalities encountered in human disease
International Consortium of Human Phenotype Terminologies (ICHPT) Standard The ICHPT provides the community with a set of terms to describe phenotypic features to be used by any terminologies to achieve interoperability between databases, in particular, to allow the linking of phenotype and genotype databases for RDs

Networking patient registries
TREAT-NMD patient registries Platform The TREAT-NMD Patient Registries is a global network of national registries that provides a unique entry point for access to rare neuromuscular disease patients worldwide

Therapeutic development
Standard operating procedures for preclinical efficacy studies Guideline Standard operating procedures for preclinical efficacy studies are a compilation of experimental protocols to measure drug efficacy in models of neuromuscular disease
Care and Trial Site Registry Platform The Care and Trial Site Registry aims to assist pharmaceutical industry and clinical investigators in deciding on clinical trial site location and in the identification of potential partners for future research projects
TREAT-NMD Advisory Committee for Therapeutics Advisory Committee TREAT-NMD Advisory is a group of experts from various origins (academic, industry drug development, patient representatives, and governmental representatives) that provide guidance on the translation of therapeutic programs in rare neuromuscular diseases

Table 3: IRDiRC Recognized Resources

Therapeutic development
IRDiRC aims to stimulate the development of 200 new therapies by 2020. TREAT-NMD has been active in confronting barriers in this space, producing resources to contribute to different phases of therapeutic development, from translation of basic research to clinical trials. Three additional resources from TREAT-NMD met IRDiRC’s requirement regarding the adequate and timely exchange of scientific and regulatory information about clinical research, and are therefore recognised as important tools towards development of new therapies. First, its ‘Standard Operating Procedures for preclinical efficacy studies’ consist of a collection of experimental protocols for the most common outcome measures used in the assessment of drug efficacy in mammalian models of rare neuromuscular diseases. A wide use of such standardised protocols would contribute to improving robustness of preclinical results that serve to justify patient trials. One of the hurdles prior to initiating a clinical trial is the identification of trial sites capable of recruitment of sufficient number of patients, and qualified personnel to provide care and experience given a specific standard. The aim of the TREAT-NMD Care and Trial Site Registry is to help industry and clinical investigators identify and select trial sites and potential partners for clinical studies in neuromuscular and neurodegenerative diseases. The TREAT-NMD Advisory Committee for Therapeutics is composed of drug development experts from academia and industry, as well as representatives from patient and scientific research centres. This committee meets twice yearly to review and provide guidance on the translation and therapeutic development path for rare neuromuscular diseases with large unmet needs. Widespread adoption by the community of such recognised resources is expected to facilitate the IRDiRC goal to enable the development of 200 new therapies by 2020.

CONCLUSION
After its first year, an assessment of the ‘IRDiRC Recognized Resources’ indicator shows that this initiative has highlighted 13 resources that focus on key IRDiRC priorities in advancing RD research: data sharing for discovery, knowledge organisation and ontologies, networking patient registries, and therapeutic development. It is expected that as more resources are highlighted and used by researchers, the pace of discovery and translation into the clinic will be further enhanced. Reciprocal recognition of resources between
international efforts with overlapping goals (e.g. Human Variome Project) will be an important step forward. Moreover, as the research community converges on and adopts recognized standards, guidelines, tools, and platforms, the resulting interoperability will enable enhanced sharing, and thus accelerate advances in research and development across all RD.

CONFLICT OF INTEREST
The authors declare no conflict of interest.

ACKNOWLEDGEMENTS
The ‘IRDiRC Recognized Resources’ initiative has been supported by members of the IRDiRC Scientific Committees. Members include Fowzan Sami Alkuraya, Diego Ardigò, Michael Bamshad, Gert-Jan Boudewijn van Ommen, Anthony Brookes, Han Brunner, Angel Carracedo, Seng Cheng, Gema Chicano, Robin Conwit, Johan den Dunnen, Xavier Estivill, Jack Goldblatt, Stephen Groft, Shuling Guo, Adam Heathfield, Bartha Maria Knoppers, Jeffrey Krischer, Milan Mack, Sandrine Mariear, Gert Matthijs, Woong-Yang Park, Samantha Parker, Ada Pitkanen, Catherine Rademaker, Pak-Chung Sham, Rumen Stefanov, Hendrik Stunnenberg, Domenica Taruscio, Josep Torrent i Farnell, Anne Zajicek, and Feng Zhang. We also acknowledge Ségolène Aymé, former Secretariat, for her assistance in launching this initiative. The work was supported by the European FP7 contract, ‘SUPPORT-IRDiRC’ (No. 305207). CPA has contributed to this work in his capacity as Chair of the IRDiRC Consortium Assembly, not in his role as Director of the National Center for Advancing Translational Sciences.

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