Orphan Drug Development Guidebook

Building Block I409

This document defines the content of the Building Block created for each identified tool, incentives, initiative or practice introduced by public bodies or used by developers to expedite drug development in Rare Diseases (RDs).

ITEM	DESCRIPTION
Building Block (BB) Title	Orphanet database
References	www.orphadata.com (formerly www.orphadata.org) Activity report(s): https://www.orpha.net/consor/cgi-bin/Education_Home.php?
Description	Orphanet is a unique resource, gathering and improving knowledge on rare diseases so as to improve the diagnosis, care and treatment of patients with rare diseases. Orphanet aims to provide high-quality information on rare diseases, and ensure equal access to knowledge for all stakeholders. Orphanet also maintains the Orphanet rare disease nomenclature (ORPHAcode), essential in improving the visibility of rare diseases in health and research information systems. Orphanet works towards meeting three main goals: 1. Improve the visibility of rare diseases in the fields of healthcare and research by maintaining the Orphanet rare disease nomenclature (ORPHAcode): providing a common language to understand each other across the rare disease field 2. Provide high-quality information on rare diseases and expertise, ensuring equal access to knowledge for all stakeholders: orientating users and actors in the field in the mass of information online.

	3. Contribute to generating knowledge on rare diseases: piecing together the parts of the puzzle to better understand rare diseases. Orphanet offers a range of freely accessible services: An inventory of rare diseases mapped with resources as OMIM, ICD10, MeSH, MedDRA, GARD and UMLS and a classification of diseases elaborated using existing published expert classifications. Diseases are also annotated with phenotypic features and frequency using HPO. An encyclopaedia of rare diseases in English, progressively translated into the other languages of the website. An inventory of orphan drugs at all stages of development. A directory of expert resources, providing information on expert clinics, medical laboratories, ongoing research projects, clinical trials, registries, networks, technological platforms and patient organisations, in the field of rare diseases, in each of the countries in Orphanet's network. An assistance-to-diagnosis tool allowing users to search by signs and symptoms. An encyclopaedia of recommendations and guidelines for emergency medical care and anaesthesia. A fortnightly newsletter, OrphaNews, which gives an overview of scientific and political current affairs in the field of rare diseases and orphan drugs, in English, French and Italian. A collection of thematic reports, the Orphanet Reports Series, focusing on overarching themes, directly downloadable from the website. A platform, Orphadata, providing high-quality datasets related to rare diseases and Orphan Drugs, in a reusable and computable format. The Orphanet Rare Disease Ontology (ORDO), a structured vocabulary for rare diseases derived from the Orphanet database, capturing relationships between diseases, genes and other relevant features. ORDO provides integrated, re-usable data for computational analysis. Orphanet and ORDO are IRDiRC Recognized Resources.

Category	Regulatory Building Block
Geographical scope	Orphanet is a multi-stakeholder, global network of 40 countries, coordinated by the Orphanet Coordinating team at the French National Institute of Health and Medical Research (INSERM) in Paris (France) List of Network Members (update 2023): https://www.orpha.net/orphacom/cahiers/docs/GB/Orphanet_Network_MB_members.pdf
Availability	Applicants developing medicines for rare diseases. Data is RD specific and available for all types of publics at the following conditions: • Products promoting the interoperabity of rare disease data on www.orphadata.com: free, open access, CC BY 4.0 1. Orphanet nomenclature and definitions, classifications and cross-references 2. Disease-gene annotations and cross-references 3. Disease-phenotype annotations 4. Epidemiological/natural history annotations 5. Orphanet Rare Disease Ontology and HPO-ORDO Ontological Module (HOOM) • Data sets requiring DTA for academia/ fee for industry: 1. Textual information 2. Catalogue of expert services/ressources 3. Orphan drug data
Scope of use	Academic researchers and the pharmaceutical industry use Orphadata datasets for research and development purposes. Examples of pharma industry use cases include the incorporation of Orphadata datasets in pre-competitive tools in order to prioritise therapies development, as well as epidemiological data to assess market size. Other use cases include datamining technology applications to generate hypotheses from Orphanet data and textual information. Epidemiological data: determination of the size of the market per geographical region. Recruitment, collaborations: identification of experts, patient organisations, registries, biobanks, research groups developing i.e. animal models, biomarkers, targets, etc, by disease/group of diseases. Pre-competitive tools: integration with other sources of data, including private pharma data, to generate hypothesis.

Stakeholders	• Funders: Public: Inserm, French Ministry of Health, European Commission, Agence de la Biomédecine, Ministries of health, universities and hospitals in the countries of the Orphanet network; Private: AFM-Téléthon, Fondation Groupama pour la Santé • Pharma companies and bio-techs using for-fee data • Academic researchers, public administrations and Patient organisations: using the open access datasets • Health professionals, patients, researchers, students, decision makers (Orphanet website users)
Enablers/ Requirements	Any particular requirement for open access datasets (see above) that are available in: • XML • JSON • Sparql EndPoint • Ontology: OWL, txt, obo (and for ORDO : http://bioportal.bioontology.org/ontologies/ORDO (OWL, CSV, RDF/XML) DTA signature for restricted access datasets (academia, public not-for profit institutions); contract (for fee) for private for-profit companies.
Output	A public website, www.orpha.net A download platform (reusable datasets) www.orphadata.com(formerly www.orphadata.org). Ontologies: ORDO and HOOM (https://www.orphadata.com/ontologies/)
Best time to apply and time window	N/A
Expert tips	PROs: – Computable structured data, manually curated, organized hierarchically (thus allowing for clustering and aggregation of data) and described with unique identifiers. Numerous cross-references with other relevant resources (gene function, pathways, compounds and targets,…) – Versioning and differentials for open access data. Documentation provided.

	– A number of channels are available to help users wishing to access and re-use Orphanet data: https://www.orphadata.com/contact/. A FAQ and user guide is also available. In addition, there is an ORDO user mailing list (https://international.orphanews.org/home.html) to deliver updates concerning the ontology. CONs: – Dataset formats are standardized and on-demand, customized datasets as a service are not yet developed (but will be) – APIs in development, not yet available. – Hands-on assistance can be necessary to make the best use of the resource (as proven by the Orphanet participation to the training courses organized by ISS for RD registries: BYOD (RD-Connect/Excelerate). Not already available as a service, but in discussion.