Drug Repurposing Guidebook

Building Block I447

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

ITEM	DESCRIPTION
Building Block (BB) Title	**How to maintain a literature archive**
References	**[1]** Shourick J et al., 2021. *Assessing rare diseases prevalence using literature quantification.* Orphanet J Rare Dis. 16:139. https://doi.org/10.1186/s13023-020-01639-7 **[2]** Frederiksen S et al., 2022. *Rare disorders have many faces: in silico characterization of rare disorder spectrum*. Orphanet J Rare Dis. 17:76. https://doi.org/10.1186/s13023-022-02217-9 **[3]** Ehrhart F et al., 2021. *A resource to explore the discovery of rare diseases and their causative genes.* Sci Data. 8:124. https://doi.org/10.1038/s41597-021-00905-y **[4]** Smith C et al., 2022. *Estimating the number of diseases – the concept of rare, ultra-rare, and hyper-rare*. iScience. 25:104698. https://doi.org/10.1016/j.isci.2022.104698 **[5]** Nguengang Wakap S et al., 2020. *Estimating cumulative point prevalence of rare diseases: analysis of the Orphanet database.* Eur J Hum Genet. 28:165-173. https://doi.org/10.1038/s41431-019-0508-0 **[6]** Masoudi-Sobhanzadeh Y et al., 2020. *Drug databases and their contributions to drug repurposing.* Genomics. 112: 1087-1095, https://doi.org/10.1016/j.ygeno.2019.06.021 **[7]** PubMed Help page https://pubmed.ncbi.nlm.nih.gov/help/ **[8]** Google Scholar Help page https://scholar.google.com/intl/en/scholar/help.html **[9]** Lens Scholar Help page https://support.lens.org/article-categories/scholar/ **[10]** Clinicaltrials.gov Help page https://www.clinicaltrials.gov/ct2/help/how-find/index **[11]** EU Clinical Trials Register https://www.clinicaltrialsregister.eu/ctr-search/search **[12]** The 5-part article *"Searching the literature for studies for a systematic review"* in Am J Orthod Dentofacial Orthop. (A Littlewood and D. Kloukos; 2019) https://pubmed.ncbi.nlm.nih.gov/31153512,31053291,30935616,30712702,30826048 **[13]** Massonnaud C et al., 2020. *Identification of the Best Semantic Expansion to Query PubMed Through Automatic Performance Assessment of Four Search Strategies on All Medical Subject Heading Descriptors: Comparative Study*. JMIR Med Inform. 8:e12799. https://doi.org/10.2196/12799

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	[14] Lacey P, 2022. *Google is goodish: An information literacy course designed to teach users why Google may not always be the best place to search for evidence.* Health Info Libr J. 39:91-95. https://onlinelibrary.wiley.com/doi/10.1111/hir.12401 **[15]** Saxena R and Kaushik J, 2022. *Referencing Made Easy: Reference Management Software.* Indian Pediatr. 59:245-9 https://www.indianpediatrics.net/mar2022/245.pdf **[16]** Wikipedia page comparing main features of reference management software https://en.wikipedia.org/wiki/Comparison_of_reference_management_software **[17]** Dos Santos Vieira B et al., 2022. *Towards FAIRification of sensitive and fragmented rare disease patient data: challenges and solutions in European reference network registries.* Orphanet J Rare Dis. 17:436. https://www.ncbi.nlm.nih.gov/pmc/articles/PMC9749345 **[18]** Abaza H et al., 2022. *Domain-Specific Common Data Elements for Rare Disease Registration:Conceptual Approach of a European Joint Initiative Toward Semantic Interoperability in Rare Disease Research.* Indian Pediatr. 59:245-9 JMIR Med Inform. 10:e32158. https://www.ncbi.nlm.nih.gov/pmc/articles/PMC9166638/
Description	Even a quick analysis of literature shows the progressive growth in scientific publishing activities disclosing properties of commercially available drugs as well as the medical management and the pathobiological mechanisms of rare diseases. Indeed, the correct identification and categorization of literature disclosing biological, epidemiological and clinical data for a given rare disease is challenging and with evolving definitions and criteria [1-6]. Thus, it is important to provide investigators with a guidance about how to effectively extract, archive, and compare the findings from biomedical literature in order to elaborate new work hypotheses for selecting candidate drugs to be repurposed in a rare disease and evaluate risks/opportunities in testing such compound(s) in models or patients. This BB provides investigators with an overview of most representative databases, search strategies, and means to archive and distribute a literature archive dedicated to a drug repurposing, proposing simple case studies and guidance about: - using four, freely available sources for biomedical publications and information being PubMed and related NCBI/EBI resources [7], Google Scholar [8], Lens Scholar [9], Clinicaltrials.gov [10], and EU Clinical Trials Register [11], comparing their specificities and (dis)advantages when searching, extracting and archiving drug repurposing & rare disease information; - How expanding search strategies for retrieving relevant literature [12-14]; - How using reference management software for archiving literature, in particular free tools such as Mendeley and Zotero [15, 16].
Category	Contact with TTO and Patents

ITEM	DESCRIPTION
Type of BB	Development practice
Geographical scope	International
Availability	It covers information resources are freely available (unless indicated otherwise, in particular for copyright reasons or subscription-based access)
Scope of use	Support to investigators involved in Drug Repurposing & Rare Disease research by avoiding duplicated efforts and improving the process for selection & (pre)clinical validation of drug candidates by: - Increasing awareness about the means to identify, use, and share relevant (pre)clinical information that is actually available in scientific literature; and - Improving the practices about the use of databases of scientific literature, the management of literature archives, and the extraction of medical hypotheses and evidences from (pre)clinical publications.
Stakeholders involved	Investigators involved in (pre)clinical research activities and data analysis; patients' organizations; professionals within academic institutions, agencies and companies that work in the fields of drug development, regulatory affairs and health policy.
Enablers/ Requirements	Previous experience in - use of internet resources for scientific research and literature; - drug and/or rare disease research, clinical, or regulatory activities.
Output	Useful knowledge for faster selection, validation, and access to new therapeutic opportunities in rare diseases taking advantage of available (pre)clinical evidences.
Best time to apply and time window	This BB is mainly applicable in the early phases of drug development, to gather all relevant information before/during the process for selection & (pre)clinical validation of drug candidates but it can support activities also in later steps, before taking any major commitment or decision when pursuing the regulatory proceedings and facilitating the access to the selected drug (acquisition, distribution, manufacturing, clinical use, safety, reimbursement).
Expert tips	PROs: - Several examples, documentation, and support for quickly applying the guidance can be found in the internet; - Compliance with open access, interoperable data and resources for promoting exchanges among investigators and European Networks (FAIRification [17, 18]). CONs: - A regular use of the described databases, search strategies, and software is needed to consolidate the knowledge and skills based on this BB;

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	- The databases and software may quickly evolve over time and some details in the BB may become incomplete/incorrect over time.