Orphan Drug Development Guidebook

Building Block I419

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	Registries for Rare Diseases
References	Orphanet: Registries & biobanks www.ncbi.nlm.nih.gov/books/NBK208609/
Description	Registries collect information on patients afflicted by a particular disease or group of diseases. By combining data on as many patients as possible, at the regional, national, European or global level, the potency of the data increases exponentially. Registries, particularly when used by many different centers, enable researchers to accrue a so-called 'critical mass' of patients which would often otherwise be impossible. Data on any rare condition is extremely precious. No single country will see a sufficient number of patients with any very rare disease to fully understand the condition, in terms of its epidemiology (e.g. how many cases exists in any given population), the range of symptoms observed, the development of the disease over time, and the likely outlook for newly-diagnosed patients, the effect in terms of safety and efficacy of therapies tested in clinical trials or in post-marketing surveillance programs. The generation of validated data regarding natural histories of rare diseases helps to define the target to address for the therapy of rare diseases, the endpoints to be matched by innovative therapies and the level of safety and efficacy reached by therapies by comparing pre and post treatment data.

Category	Development Practices Building Block
Geographical scope	International
Availability	Applicants developing medicines for rare diseases.
Scope of use	– Natural Histories of RD: By collecting data over a long period of time – Epidemiology of RD: how the disease is caused and the incidence on a certain population – Assessment of disease threats and planning of health services: analysis of quality of life – Assessment of efficacy and efficiency of diagnosis – Assessment of the safety, efficacy and efficiency of treatments – Choice of endpoints to be used in clinical trials on different population and at different disease conditions. – Post Marketing surveillance – Genotype-phenotype correlation
Stakeholders	• Healthcare professionals • Pharmaceutical industries • Patient organizations • EC representatives
Enablers/ Requirements	Enablers: ERNs representatives, Patients Organization and Pharmaceutical Industries, EC representatives for Information and consultantships. Requirements: • Inform (e.g. announcement of guidance on registries); • Consult (written – e.g. surveys); • Consult and involve (direct interactions – e.g. stakeholder meetings, workshops, stakeholder conferences); • Cooperate / participate (direct interactions - e.g. technical expert groups
Output	Report on the mapping of registries for RD, generation of recommendation regarding the need of unified registries.

Best time to apply and time window	The tool has its best use at the start of development.
Expert tips	No