Orphan Drug Development Guidebook

Building Block I427

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	New-born screening programs for rare diseases
References	www.cdc.gov/newbornscreening/ www.nichd.nih.gov/health/topics/newborn/conditioninfo/purpose
Description	The purpose of NBS is to detect potentially fatal or disabling conditions in newborns as early as possible and possibly before onset of symptoms. Such detection allows the early treatment which may significantly modify the natural history of the disease and potentially prevent developmental delays, physical disabilities and eventually death. The identification of rare diseases before the onset and the clinical evidence gives to the physician the possibility of choosing the most adequate treatment and to the patient the possibility to benefit of the treatment before the onset of irreversible defects. Regarding the development of new drugs, the possibility of studying pre-symptomatic patient might facilitate the understanding of the real efficacy of innovative therapies in modifying the natural history of diseases.
Category	Developmental Resources Building Block
Geographical scope	International

Availability	Applicants developing medicines for rare diseases.
Scope of use	– Identification of rare diseases at early stage, potentially at the pre-symptomatic stage – Epidemiology of RD: how the disease is caused and the incidence on a certain population – Prevention 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 – Generation of awareness programs on RD for the early identification and treatment of RD – Generation of new technologies for NBS methods
Stakeholders	• Healthcare professionals, • Pharmaceutical industries/drug developers • Patient organizations, • Policy makers
Enablers / Requirements	Enablers: ERNs representatives, Patients Organization and Pharmaceutical Industries, BoMS Requirements: • Inform (e.g. announcement of guidance on NBS); • 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 the situation of NBS for RD, generation of recommendation regarding the need of apply NBS in an extended manner.
Best time to apply	The tool has its use as early as possible at disease knowledge phase.

and time window	
Expert tips	PROs: – Early diagnosis, early treatment, generation of data about efficacy of therapies, more adequate choice of the treatment is available, better follow up with supportive therapies, prevention of life-threatening events and casualties. – Generation of data regarding epidemiology and prevalence of RDs. CONs: – Increase of diagnostic cost for the NHS, increase of cost due to the treatments applied to pre-symptomatic and early identified patients.