Pilot evaluation Steering group UNL-NFU-NWO-UKB – Elsevier

This document assesses to what extent the agreed principles for the specific pilot service have been met. This document goes with the pilot evaluation document and is used by the steering group to sign-off on the pilot's evaluation.

Name Pilot/Service: Rare Disease Analytics

Short description of the project:

Introduction

Rare diseases are, despite its name, very frequent: about 8000 rare diseases hit approximately 30 million people in Europe alone (1.5 million in the Netherlands). Though there is quite some research done into many rare diseases, their findings are hidden in the enormous number of medical publications. Their metadata is usually not adequately indexed by search engines like PubMed or Embase and therefore difficult to discover.
The Dutch Ministry of Health assigned about 300 expertise centers for these rare diseases, but their research and outcome (articles) overview are often incomplete or take a lot of time to compose manually. By refining the indexing in Scopus based on an Erasmus MC prototype the metadata of these articles will be improved and linked to the relevant centers.

The aim of this pilot is to provide open access to insights (articles, metadata, other information) about rare (or orphan) disease research activities in the Netherlands to researchers, institutions, patients, funders and other stakeholders in health care, stimulating further research (patient data) to rare diseases and contributing to public knowledge.

1. (a) Participating institutions Participation in the Professional Services is at each Institution's sole discretion and a pilot shall only commence if there is a minimum participation by at least three Institutions	Evaluation To what extent did we deliver as anticipated	Evidence and Comments listed in the framework document
Are at least 3 institutions involved in the pilot?	**3 institutions signed the SoW (AUMC, UMCG and Erasmus MC) + endorsement by NFU (public statement)**	The project has been executed with help and support of most NFU members and the VSOP.
Evidence of how and when other institutions can join	**Other MCs are still welcome to join if they want to**	2nd and 3rd tier hospitals could join as well, as well as independent research institutions in medicine and patient organizations.

1. (b) Interoperability and vendor neutrality Elsevier shall use all reasonable efforts to ensure that the Professional Services are interoperable, both on the input (uploaded) and output side (created)	Evaluation To what extent did we deliver as anticipated	Evidence and Comments listed in the framework document
Use of open identifier systems	**Use of orpha.net as planned**	Orpha (see orpha.net) and DOI
Use of standardized metadata schemas	N/A	
Existence of a well-documented API and open data-dump function	**An API was not used. We did share a table of DOI-ORPHA relations, but the CRIS system could not handle such a large classification**	If an API is required, it will be one using standard technology
Ability to export data in a variety of formats	**All that is needed are lists connecting Orpha codes with DOIs.**	Most likely all that is needed will be lists connecting Orpha codes with DOIs.
Ability for other commercial parties to join	**Each member is free to share its DOI-Orpha codes links**	Members can share their Orpha codes per DOI with anyone.
2. Transparency, inclusion, and collaboration The Services and resulting Deliverables are aimed to make science and research more transparent, efficient, inclusive, openly and freely accessible, and collaborative.	Evaluation To what extent did we deliver as anticipated	Evidence and Comments listed in the framework document
Provenance on how and where metadata was derived	**We used a proprietary NLP-based algorithm TERMite (of which the principle has be explained at 2 conferences) to match Orpha codes and DOIs.**	The current plan is to use a proprietary NLP-based algorithm (of which the principle will be explained in exact wording) to match Orpha codes and DOIs.
Descriptions of workflows that result in indicators, metrics and/or other relevant outcomes will be open and transparent. These will demonstrate, for example calculation steps, search strings used to define entities, etc.	**The dashboard has been created as planned. Experts tested data and provided desired specs for the dashboard, as well as feedback on versions.**	The dashboard will be created with a representation of the community of rare disease experts. (about 50% of academic researchers in UMC's are likely involved with rare disease research.) They test data and

		provide desired specs for the dashboard
Description of the services used to create (meta)data	**No change vs. Original plan**	NLP-based algorithm (of which the principle will be described/published in relevant conference proceedings) creates search queries for each Orpha code. These queries will run over Scopus data and generate lists of DOIs per Orpha code.
Insights and lessons published with Open Access license	NA	We plan to publish methodology and process in two separate scientific papers in OA

Will the pilot contribute to Open Science?	**The pilot brought the expected outcomes (see evaluation document)**	The Creation of an open system that provide insights in Rare Disease research progress will create
		- efficiencies for the evaluation processes already in place
		- visibility of outputs and outcomes per disease or disease cluster for:
		  o research groups
		  o institutions
		  o funders
		  o Governments
		  o Patient Groups
		- visibility of research efforts and experts for patients
		- visibility of developments, trends and experts for:
		  o pharmaceutical companies
		  o health insurers
		- This project will support Evidence Based Medicine, (medical practice or care that emphasizes the practical application of the findings of the best available current research)

Demonstration of connection to non-Elsevier products	**No API was requested by the participating institutions**	DOIs are used by all vendors in research eco system. The Orpha codes are publicly accessible. If an API is required, we will provide one that uses standard technology.
3. Access to research data and metadata Elsevier will give enduring access during the Term to all (research) data, including metadata, analytics and information	Evaluation To what extent did we deliver as anticipated	Evidence Comments listed in the framework document
Describe the ownership / licensing of data made as part of the service	**The dashboard is publicly available from the epdos.nl website: https://epdos.nl/rarediseasemonitor/**	The connection between Orpha codes and Dutch DOIs will be made available on a public site and to the individual members.
Describe how access (institutional and / or public) to the data will be set-up during the term; this section will also indicate cases where certain data is not publicly access.	**The dashboard is publicly available from the epdos.nl website: https://epdos.nl/rarediseasemonitor/**	NA
4. Data portability Institution shall be entitled to transfer the data provided, uploaded, or created to its own or to a third-party host environment	Evaluation To what extent did we deliver as anticipated	Evidence Comments listed in the framework document
Evidence on how data transfer is possible	**List of documents were provided to institutions who requested them**	Lists of Orpha-DOI combinations will be provided to the institutions; format will be agreed by the parties. Unfortunately the classification was too large for CRIS systems, for now.
How can an institution withdraw data?	N/A	
5. Intellectual property	Evaluation To what extent did we deliver as anticipated	Evidence Comments listed in the framework document
Details on IP related to data provided, created, or enriched	**The service uses a proprietary NLP-based algorithm (TERMite) to match Orpha codes with DOIs. The principles of the algorithm will be**	The service will use a proprietary NLP-based algorithm to match Orpha codes with DOIs. The principles of the algorithm will be described/published in two

	described/published in two conference papers and an upcoming journal article.	conference papers and an upcoming journal article.
6. Additional considerations	Evaluation To what extent did we deliver as anticipated	Evidence Comments listed in the framework document
What processes will be put in place to evaluate the service during and at the end of pilot	**The service was as planned a co-creation with NFU and senior NFU officials supervised the project. A review process enabled incremental improvements throughout the project.**	The service will be a co-creation with NFU and will be supervised by senior NFU officials. In alignment with the participating institutions, a review process will be setup to test the outcomes of the service.
Terms of use of the deliverables during and after contract period	**No change.**	The deliverables of this pilot are stated in the SoW. If participating institutions so wish, the service is available until the end of 2024. All data provided to the institutions remains with the institutions also after the end of this agreement.
Pilot project team	**The service has been a co-creation with NFU and was supervised by senior NFU officials**	The service will be a co-creation with NFU and will be supervised by senior NFU officials