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Facility for the visualisation of the hierarchical organisation of the ORPHA nomenclature

DELIVERABLE 4.7

December 2020

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Table des matières

Introduction

Within the Subtask 4.5.3 of the project the web-based applications development by Orphanet was foreseen. The aim was to facilitate the informatic access to nomenclature data and allow flexible implementation into the various IT systems in use in the different countries and/or settings.
An API https://api.orphacode.org/ , was released as a beta version in December 2019 (API for visualisation of Orphanet Nomenclature Deliverable 4.2) and then the official release and upgrade was made in June 2020 (Facility for ORPHA nomenclature correspondence, Deliverable 4.5). The Orphanet Data visualisation tool https://dataviz.orphacode.org/ is delivered in December 2020.

Orphanet Data visualisation Tool December 2020 release

The tool is accessible here: https://dataviz.orphacode.org/ . All interrogations exploit the above-mentioned Orphanet API also released within this project, showing at the same time the nice exploitation potential of the Orphanet API. This tool allows to search for the clinical entities (groups of disorders, disorders, sub-types) that are present in the nomenclature pack (here) in all the API languages (fig.1).

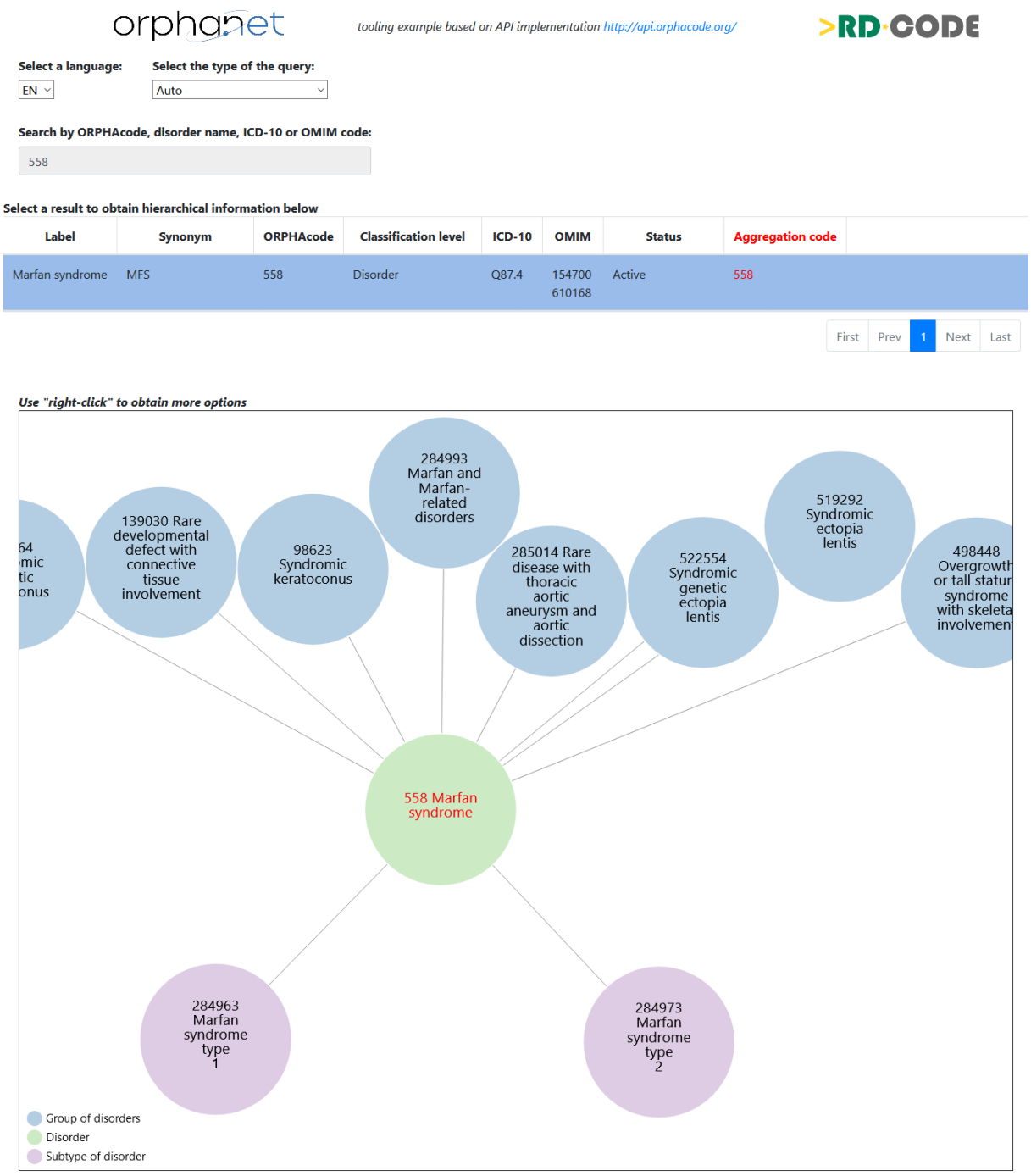

Figure 1 Tool homepage with search results for ORPHAcode 558 Marfan Syndrome

Search/Queries can be done by Preferred term; Synonym, both, ORPHAcode, ICD-10 code and OMIM number (fig.2). The search by label is error tolerant so that as an example “hircsprung or hirshprung” should work.

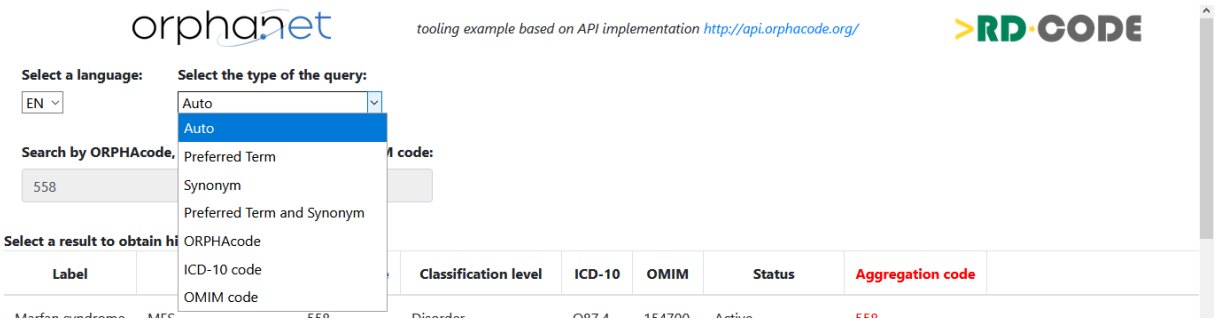

Figure 2 Search/Query Selection possibilities in the tool.

The result list provides for each entity the label, the Synonym(s), the ORPHAcode, the classification level, the ICD10 code(s), the OMIM number(s), the status (Active/inactive) as well as the aggregation code (fig.3).

Then you select the results of interest for you in the list provided (fig.3)

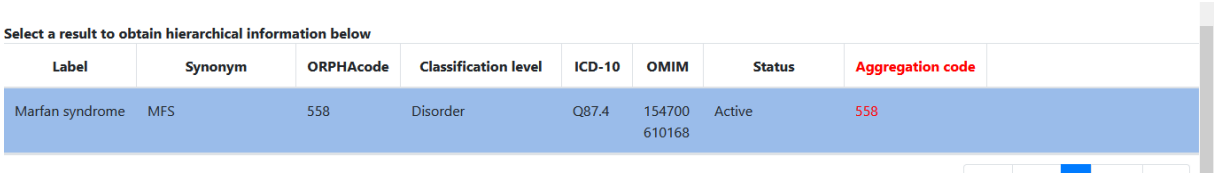

Figure 3 Selection of the relevant result

This will generate the view as indicated in fig 4. At the center the Disorder (color code light green), above the groups to which it belongs (color code light blue) and below the subtypes if relevant (color code light pink).

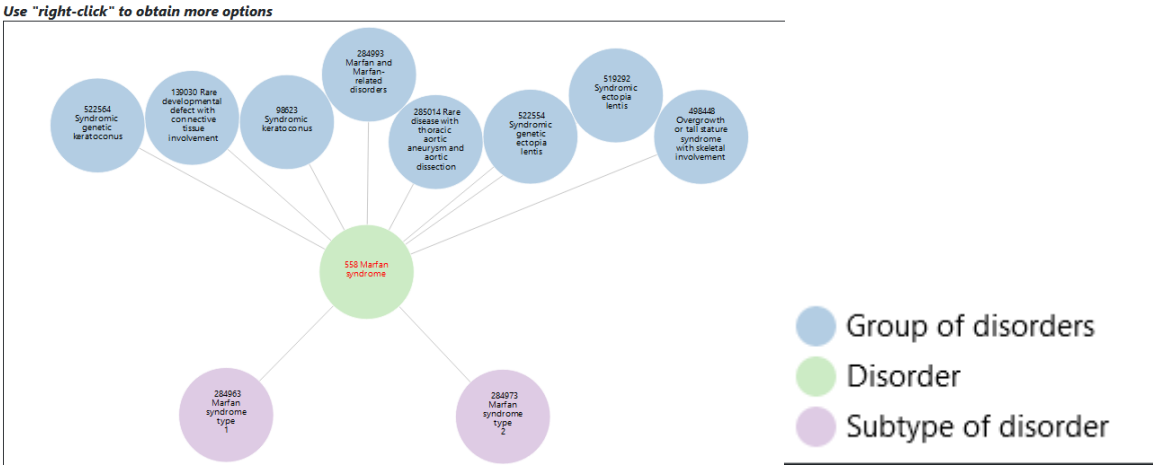

Figure 4 Visualisation of the Orphanet Classification for ORPHAcode 558, Marfan syndrome and color code for classification level.

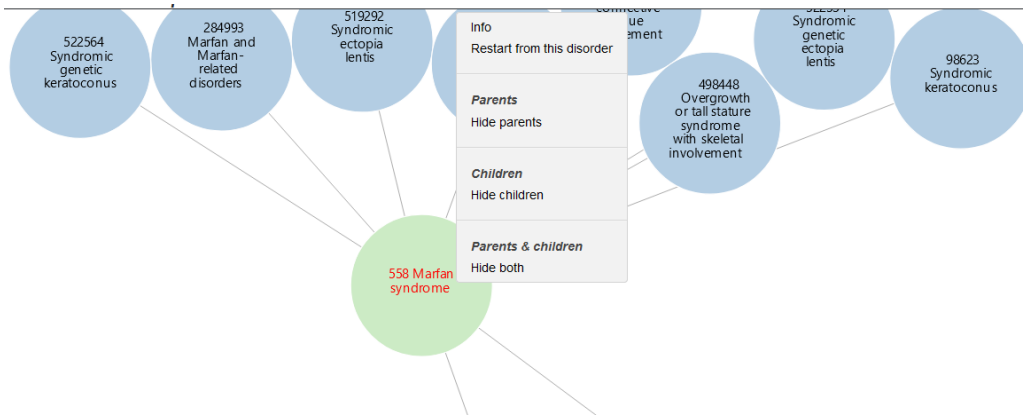

Figure 5 Menu allowing to obtain more info, available for each "bubble".

In fig.6 it is shown the new visual representation when asking to show "descendants" of ORPHAcode ORPHA:522564Syndromic Genetic keratoconus, one of the group of disorders Marfan Syndrome belongs to. Results obtained when querying for ORPHAcode 581 Mucopolysaccharidosis type 3 are indicated in fig.7 .

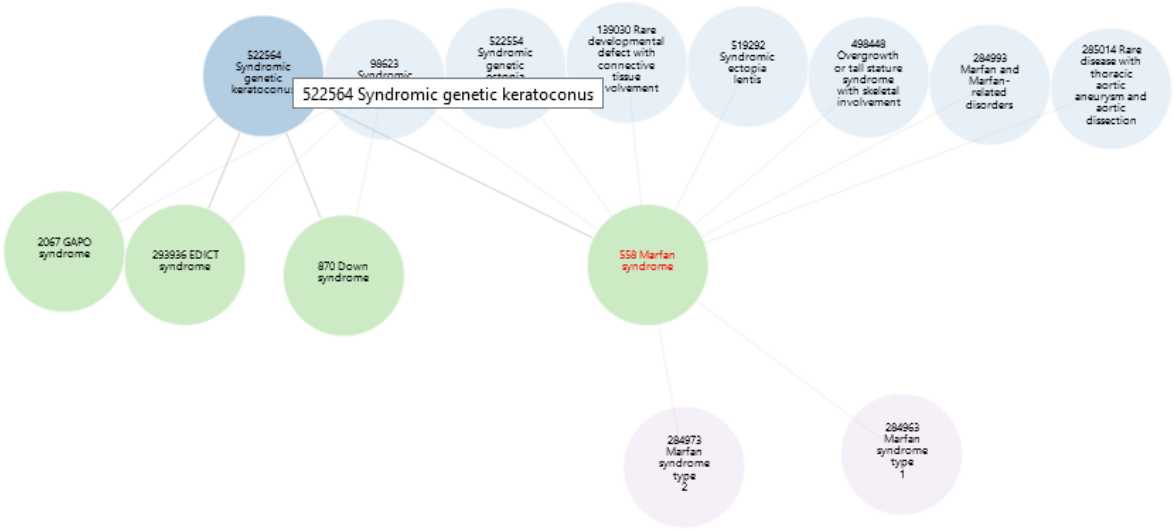

Figure 6 Right click to show "descendants" of ORPHAcode 522564, Syndromic Genetic keratoconus

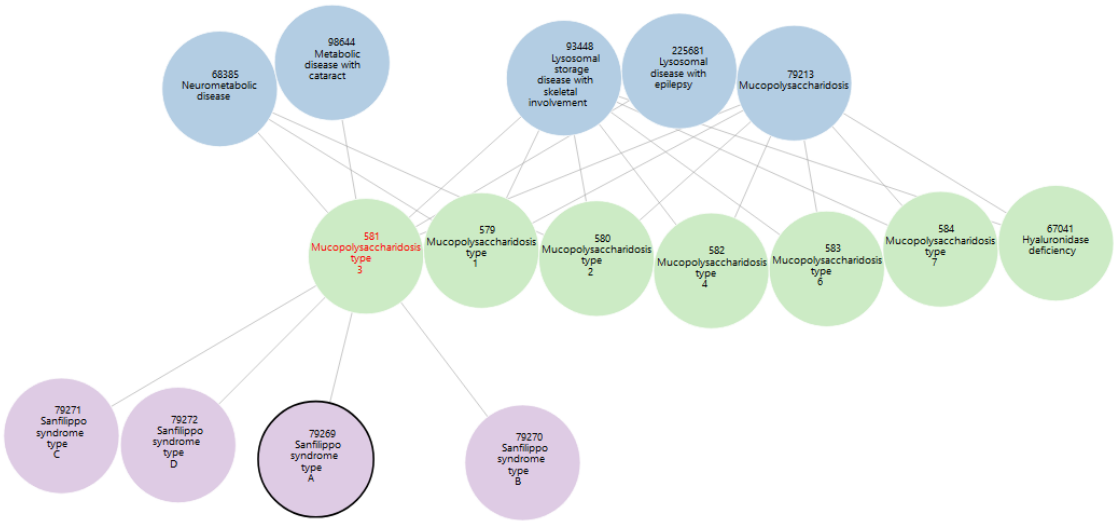

Figure 7 Results for query for ORPHAcode 581, Mucopolysaccharidosis type 3

Code avaibility

The tool code is released in a dedicated GitHub repository under RDCode : https://github.com/orphanet-rare-diseases-issues/dataviz_poc1

Therefore, this web application could be deployed in several environment or web server at glance. As it uses mainly Javascript code to perform operations, it's a lightweight webapp. However, an internet connexion is mandatory as the tool exploit the accessible online API.

The graphical representation use D3 javascript library under licence: https://github.com/orphanet-rare-diseases-issues/dataviz_poc1/blob/main/lib/LICENSE.D3 .