

From FAIR4Health Project to 1+MG Initiative: A Spain – Italy Collaboration

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Abstract. Results of two major projects funded by the European Union are taken into consideration: Fair4Health regarding the possibility of sharing clinical data in various environments applying FAIR principles and 1+Million Genome for the in-depth study of the human genome in Europe. Specifically, the Gaslini hospital plans to move on both areas joining the Hospital on FHIR initiative matured within the fair4health project and also collaborate with other Italian healthcare facilities through the implementation of a Proof of Concept (PoC) in the 1+MG. The aim of this short paper is to evaluate the applicability of some of the tools of the fair4health project to the Gaslini infrastructure to facilitate its participation in the PoC. One of the aims is also to prove the possibility of reuse the results of well-performed European funded projects to boost routine research in qualified healthcare facilities.

Keywords. FAIR, genome, data sharing, FHIR, translational health informatics.

1. Introduction

In recent years, it has become clear the potential impact and importance of the application of FAIR Principles [1] under the paradigm of open science and of important impact in the health domain. The interest in applying FAIR principles in health care is demonstrated by the development of many research projects funded by the European Union. Among them, one of the most interesting in recent years has been FAIR4Health [2], whose main objective is to make clinical data from different European sites available for to facilitate the European health research community's common use of such data through the application of the FAIR principles. Another very promising line of research in this decade is human genomics. In this area, one of the most relevant recent projects and of a very relevant strategic scale is 1+Million Genome (1+MG) [3], launched in 2018, which was created with the aim of enabling secure access to genomics and corresponding clinical data across Europe to improve research, personalized healthcare, and healthcare policymaking. The Gaslini Pediatric Scientific Hospitalization and Care Institute (IRCCS) plans to move on both areas joining the Hospital On FHIR initiative [4] based on the tools [5] and the HL7 FHIR4FAIR FHIR Implementation Guide [6] matured

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within the fair4health project and also to participate in collaboration with other Italian healthcare facilities through the implementation of a Proof of Concept (PoC) in the 1+MG. The aim of this poster is to formulate the applicability of some of the tools of the FAIR4Health project on the Gaslini infrastructure to facilitate its participation in the PoC.

2. Methods

The main outcome of FAIR4Health has been to develop a set of open software tools, among them the ones we want to evaluate useful for our paper are the Data Curation Tool and the Data Privacy Tool, based on the HL7 FHIR standard. The first tool is a standalone desktop application that runs on major operating systems. This application connects to a FHIR endpoint in order to map local existing clinical data into hl7fhir resources. It is possible to select the FHIR profile and to perform mapping operation between the clinical data of the Hospital datasets with the FHIR resources. The dataset and its metadata are curated and validated and then the data privacy tool is used applying de-identification algorithms [7]. For participation in the PoC mentioned above one of the components used by other European groups within 1+MG initiative is Beacon [8], a standard and a service provided by the GPAP (Genome-phenome Analysis Platform) along with its Data Discovery functionality. It allows users to query a controlled access resource and it also allows queries based on the presence of a particular allele at a certain genomic locus [9]. Along with Beacon they used the GA4GH standard Phenopackets [10], a phenotypic data exchange protocol mapping HL7 FHIR to support exchange of phenotypic information.

3. Results

An analysis of the set of information that are necessary for their correct use is underway within a real Italian hospital environment. The authors try to insert these pipelines in the interoperability flow already present in the hospital taking into account all the various levels, physical, semantic and process. Specifically, most of the research documentation is available in the HL7 CDA format and the authors intend to set up elements and criteria for the correct use of this already structured data, tacking account the correct profiling of FHIR resources, both mapping with Phenopackets as the fulfilment of FAIR principles HL7 FHIR4FAIR FHIR IG-based.

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