SPHN Data Coordination Centre Seminar and Training Series featuring
international experts in the area of semantic interoperability

Date: Friday, 14th February, 2020; 10:00 – 12:00
Room: Pharmacenter, Hörsaal 2, Klingelbergstrasse 70, Basel
Contact: dcc@sib.swiss and/or register

10.00-11.00 Prof. Melissa A. Haendel
Director of the Center for Data to Health, Oregon Health & Science University

Global Alliance for Genomics and Health (GA4GH) and Phenopackets

The Global Alliance for Genomics and Health (GA4GH) is an international, non-profit alliance formed in 2013 to accelerate the potential of research and medicine to advance human health.

Bringing together 500+ leading organizations working in healthcare, research, patient advocacy, life science, and information technology, the GA4GH community is working together to create frameworks and standards to enable the responsible, voluntary, and secure sharing of genomic and health-related data. Within GA4GH Melissa Haendel is the Lead of the GA4GH Clinical & Phenotypic Data Capture Work Stream. In her talk she will present the work on the Phenopacket Schema an open standard for sharing disease and phenotype information to improve our ability to understand, diagnose, and treat both rare and common diseases. A Phenopacket links detailed phenotype descriptions with disease, patient, and genetic information, enabling clinicians, biologists, and disease and drug researchers to build more complete models of disease. The standard is designed to encourage wide adoption and synergy between the people, organizations and systems that comprise the joint effort to address human disease and biological understanding.

11.00-12.00 Prof. Harold Solbrig
Professor, Johns Hopkins University, Baltimore

FHIR, RDF, ShEx, and JSON-LD: Synergies and opportunities for SPHN

HL7 FHIR has become the de-facto standard for the exchange of clinical information. Its world-wide adoption by major healthcare providers, EMR vendors, clinical researchers and IT giants such as Apple, Amazon and Google pretty much guarantees that FHIR will be *the* model of healthcare data interchange for the foreseeable future.

The FHIR standard specifies both XML and JSON interchange formats, but also includes a specification for the representation and exchange of healthcare data in RDF. The FHIR RDF specification defines a catalogue of standard FHIR URI's that can be viewed as the equivalent of a "Dublin Core" for clinical data. The catalogue is available in both OWL and as a (draft) *schema.org* extension. FHIR RDF includes a library of Shape Expression (ShEx) schemas that can be used to validate, assemble and *transform* FHIR RDF to and from other representations. The FHIR RDF working group is investigating the use the recently published JSON Linked Data (JSON-LD) 1.1 standard as a way of combining FHIR JSON, RDF and associated semantics as single, cohesive package. Prof. Solbrig will provide a brief introduction to HL7 FHIR standard, followed by a description of the RDF specification, the FHIR URI catalogue, the ShEx RDF Schema and will then provide an update on and summary of the nascent JSON-LD FHIR representation.

For those attending the presentation in the Hörsaal 2 - followed by a buffet lunch – please register by sending a mail to: dcc@sib.swiss (seats are limited)