

Cancer Cytogenetics Beacon



This project is based on the premises that:

- cytogenetic aberration data (e.g. chromosomal translocations) represent an important set of molecular markers, especially in hematologic malignancies but also in developmental diseases
- a large body of such data exists, but is mostly difficult to access and or search, systematically
- cytogenetic annotations can be converted into modern variant annotation formats, such as being developed by the Genomic Knowledge Standards workstream (GA4GH::GKS) of the Global Alliance for Genomics and Health (GA4GH)
- an implementation to access genome variant data is the Beacon API

The project will entail

- identification and retrieval of (molecular-)cytogenetic reference datasets
- developing a cytogenetic => variant objects parsing software (based on some pre-existing code examples)
- implementation/adaptation of a GA4GH Beacon instance (e.g. from our Beacon+ resource)

The project can be adjusted to a timeframe between 6-12 months, depending on the host institution's requirement.

Participation in additional projects is possible/probable. Please contact me for details.

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