

Insights into InSiGHT and the HVP experience.

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The International Society of Gastrointestinal Hereditary Tumours (InSiGHT) is an interdisciplinary, scientific organization whose mission is to improve the quality of care of patients and their families with conditions resulting in hereditary gastrointestinal (GI) tumours. One of the many initiatives which can achieve this goal is to identify, curate and classify all of the variants which cause hereditary GI tumours. Therefore, collaboration between the Human Variome Project (HVP) and InSiGHT was a natural course of action that would benefit all stakeholders. It was agreed upon in the summer of 2007 that such a collaboration would be one of the pilot studies of the HVP. The overall aims of which would be: (1) to pilot systems to register all variants in genes predisposing to GI cancer; (2) to create a scalable platform that could be utilized by all other genes across the genome; and (3) to develop country specific nodes that are capable of being integrated. At the beginning of the partnership, InSiGHT had an official mutation database that was created through data submitted by scientists and clinicians. There were also two other databases in existence at the time, which had been created by members of InSiGHT but curated independently and run from autonomous websites. These were the Mismatch Repair Genes Variant Database (<http://www.med.mun.ca/MMRvariants/>) and the Mismatch Repair Gene Unclassified Variants Database (www.mmruv.info). For ease of use and for solidarity purposes these databases were merged in January, 2009 into a single entity and into the Leiden Open Variation Database (LOVD) platform. This amalgamated database is now housed on the InSiGHT website (<http://www.insight-group.org/mutations/>) or can be accessed directly through LOVD (http://chromium.liacs.nl/LOVD2/colon_cancer/home.php). Since this merger there have been numerous "data dumps" from different countries into the InSiGHT database. We are not just interested in collating variants but are also working towards providing better methods of text searching for variant data in the literature; annotating and evaluating functional assay information; creating a data capture system for phenotypic data; creating algorithms to determine pathogenicity; and establish privacy and confidentiality guidelines.