National Human Genome Research Institute

GENOMIC HEALTHCARE BRANCH (GHB)

NHGRI Family Health History Tool Meeting Tool Descriptions

My Family Health Portrait

My Family Health Portrait (MFHP) is a Web-based tool for collecting, organizing, and storing a record of one's family health history. It also offers disease risk estimation calculation for colorectal cancer and diabetes. The current version (3.x) was created by NHGRI along with the U.S. Surgeon General's Family History Initiative. Privacy is provided by keeping all data with the user. All processing and data storage is done on the user's device. Users may share the information with family members or doctors by printing out a chart or a pedigree drawing, or by sharing the XML data file, either directly on media or via cloud-based personal health records. This tool is unique in that it is non-commercial, NIH-supported, and client-based: no personal information passes through government servers. In addition, it is "open source." Anyone can download or fork code for more development. MFHP is compliant with HL-7, LOINC, SNOWMED-CT standards.

Myriad

Hereditary Cancer Quiz – A quick, 30 second, on-line quiz that helps patients and providers quickly identify who may be appropriate for hereditary cancer testing. The user is provided with a robust result detailing what societal criteria they have met, a patient education video, links to find a provider near them and additional online educational resources.

Family History Tool – An online tool that helps patients and providers identify who is appropriate for hereditary cancer testing based on the patient's personal and family history of cancer. The result includes a full picture of the patient's personal and family history and a 4 generation pedigree. Patients can use this application at home to self-identify for hereditary cancer testing and be more prepared to discuss their risk with their provider. Providers can use this tool in their practice to help easily identify patients, draw a pedigree and place an order for hereditary cancer testing.

ZibdyHealth- Family Medical History

ZibdyHealth allows individuals to create a private family network, in which information on disease, treatment responses, and side effects can easily be shared thus creating "***Virtual Pharmacogenomics"***. This network based approach allows family members to each share their personal medical history to compile a more accurate family history than any one person could "remember" on their own, which can then be analyzed to assess health risks. ZibdyHealth has also been designed to compile other high

quality health data in a structured manner, from barcode scanning of medications, to import of data directly from electronic health records. ZibdyHealth is aware that not all individuals are willing to share medical history with even their closest relatives, and that sharing is wrought with ethical and legal issues. By putting the information in the hands of the user, and providing multiple layers of privacy control, ZibdyHealth empowers the user to share their data as they see fit.

Health Heritage

Health Heritage (HH), a consumer facing web based application enables patients and family members to automatically extract and update clinical EMR data, share and maintain it to create electronic family histories. HH's analytic validity outperformed PCP's in completeness of family histories [Public Health Genomics 2010; 13:477–491]. It establishes a "living legacy" for children and future generations. HH's risk engine provides evidence-based recommendations to users and providers. Clinical validation demonstrated strong agreement (97%) between HH's referral for genetic evaluation versus an expert genetics team. [Familial Cancer 2015; 15:331-339]. HH went live in 2014 within the NorthShore University HealthSystem. At NorthShore, previously unrecognized high-risk patients have independently pursued genetic evaluation following a HH recommendation (clinical utility). Payers accepted HH reports as justification for genetic testing. HH was recently acquired by NantHealth, a precision medicine company. It will be used in Moonshot 2020, other research and biobanking projects. www.HealthHeritage.org, www.NantHealth.com and www.cancermoonshot2020.org.

Progeny's Family History Questionnaire

Progeny's Family History Questionnaire (FHQ) allows patients to complete their family history before the clinic visit. Questionnaires can be accessed securely via an email link, at a kiosk or tablet in the waiting room, or from a link on your website or patient portal. Pedigrees are generated from patient-entered data and can be accessed, analyzed, and modified instantly. Clinicians can enter pedigrees directly. Progeny includes integrated hereditary cancer risk assessment models including Bayes Mendel (BRCAPRO etc…) and Tyrer-Cuzick, with additional models to be added soon. Custom queries and spreadsheets can be saved, run and exported to excel or other file formats. Pedigrees can be automatically integrated with the EMR as either a hyperlink displaying a live view of the pedigree, or as a time-stamped PDF. Progeny can host your database on a secure and private cloud server. Ready-to-use templates are available to get users started quickly. Learn more at http://progenygenetics.com

CRA Health (Formerly Hughes RiskApps)

CRA Health (Formerly Hughes RiskApps) was designed to decrease physician workload and increase quality of care by allowing direct data entry by patients and by using Clinical Decision Support (CDS) to help physicians choose the best management for their patients. At the primary care level: Patients enter their family history via the web or a Tablet, the major risk models and guidelines for cancer are run and the system identifies which patients require genetic testing and/or more intensive screening. Letters are generated to the patients and their providers, simplifying compliance. At the risk clinic level: CRA Risk Clinic allows genetics professionals to manage the highest risk individuals with pedigree drawing, CDS, genetic test tracking and document generation. The system uses the HL7 standards to interoperate with the Bayes Mendel WebService, which always uses the most up to date version of each risk model).

MeTree

MeTree's purpose is to help identify patients who meet evidence-based criteria for disease prevention/risk management strategies that differ from population-based screening and to facilitate implementation of appropriate strategies. To overcome barriers to collecting, synthesizing, and acting

on family health history (FHH) derived risk information at the point of care, MeTree takes the following approaches. 1) Improve accuracy and completeness of risk information. MeTree is patient-facing with embedded education so patients have the time and knowledge to gather accurate and complete information prior to their appointment. 2) Promote shared decision making and patient engagement. MeTree provides real time guidance to the patient about their risk, what to discuss with their doctor, and pros and cons of recommended risk management. 3) Encourages uptake of clinical guidelines. MeTree provides tailored clinical decision support to physicians at the point of care with just in-time embedded education. 4) Optimize clinical workflow through a SMART-FHIR interface.

Family Healthware™ Risk Assessment

Family Healthware from Sanitas, Inc. is an FDA 510(k)-cleared web-based software application that assesses familial risk for six diseases (coronary heart disease, stroke, diabetes, breast cancer, ovarian cancer, and colorectal cancer) and provides a prevention plan with personalized recommendations for lifestyle changes and screening. Originally developed by the CDC, it analyzes health data from consumers and their biological relatives to identify the risks for acquiring major illnesses and provides a personalized risk assessment based on health data, lifestyle data, and family health history. Family Healthware deploys patented Familial Risk Stratification (FRS) algorithms to reliably generate a personal health risk score. It is intended to enable consumers to utilize family genomic data to increase disease risk perceptions and improve chronic disease management. More info at www.FamilyHealthware.com.

Proband

Proband is a free iPad application designed to enable genetic counselors and clinicians to quickly and efficiently capture a patient's genetic family history during the clinical encounter. Users create the pedigree using a series of gestures similar to drawing. All data are available in a structured format, with diagnoses from ICD-10 and the Human Phenotype Ontology. Completed pedigrees can be exported as images to multiple formats, or as data to an XML file. The Department of Biomedical and Health Informatics at The Children's Hospital of Philadelphia developed the app in collaboration with genetic counselors in multiple clinics. The NHGRI Clinical Sequencing Exploratory Research Program initially funded development; continued development is funded by CHOP. Server software called Proband Connect (currently in pilot testing) deploys behind a Hospital's firewall and enables integration of pedigree data into the electronic medical record as well as sharing between counselors. Proband has over 2500 downloads.

CancerGene Connect

CancerGene Connect (CGC) is among the original family history and risk assessment tools. CGC uses a patient-facing questionnaire to collect family structure and personal and family health history. The questionnaire utilized within CGC underwent rigorous evaluation to ensure accuracy and reliability of patient reporting[1]. The CGC platform was built to allow flexible integration with a variety of systems. Family history such as cancer diagnoses and other clinical findings are collected in a patient-centered lexicon that is mappable to clinic and billing codes. The questionnaire consists primarily of structured data which can be queried by the provider through the internal database, but is flexible enough to also allow for collection of unstructured data. Personal and family history is customizable and can be customized for clinical uses outside of cancer. End to end, the CGC program was designed to increase clinical accuracy and efficiency while minimizing clinician time and effort.

1) Pritzlaff, M., et al. An Internal Performance Assessment of CancerGene Connect: An Electronic Tool to Streamline and Improve the Genetic Counseling Process. *J Genet Counsel* (2014) 23:1034-1044

VICKY (VIrtual Counselor for Knowing Your Family History)

VICKY, a "virtual counselor", is a tool developed to overcome the literacy-related challenges in computerized family health history collection. VICKY is a Relational Agent (RA), a computer-animated character that simulates face-to-face conversation with patients. An early prototype, tested with an underserved patient population, demonstrated greater usability compared to My Family Health Portrait (Wang et al., 2015). VICKY is a web-based, patient-facing tool that collects family history on first- and second-degree relatives for 20 conditions. The tool generates a pedigree chart that users may print and share with others, and counsels users to share family history information with health providers. A Spanish language version of VICKY will be finalized later this year. VICKY can be deployed in waiting rooms or used at home, and can export HL7 FHIR data to EHR systems and risk assessment/clinical decision support tools. A trial testing the efficacy of VICKY is in progress.

CancerIQ

CancerIQ provides a suite of patient engagement tools and productivity solutions to make it easy for practices to start, run, and grow their clinical cancer genetics programs. We gather family history from patients using language they understand, provide educational content at their convenience, and make it available on the devices they already use. Patients can complete questionnaires unassisted at home, or in clinic prior to the encounter. This streamlines the workflow for genetics providers, who can use CancerIQ review and enhance pedigrees, calculate risk scores, select tests and medical management plans, and generate documentation automatically. We currently provide clinical decision support for hereditary breast, ovarian, and colon syndromes based on NCCN guidelines, and will expand to all other cancer syndromes by Q4 2016.