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RESEARCH ARTICLE

Development of a computer-based tool to obtain a family health history in Vietnam

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ABSTRACT

Background: Family health history (FHH) is central to human genomic profiling construction; however, there is no protocol for documenting FHH in a pedigree format in Vietnam.
Aim: A "Gia Su Suc Khoe" (GSSK) tool was developed to create a user-friendly interface for collecting FHH and offering diseases' risk assessment.
Results: A tool was described (https://giasusuckhoe.vn/) with good feedback from genetic counselors and family-medicine doctors. Among 20 surveys, 100% of respondents noted that the report accurately reflected their FHH and were satisfied with the tool's display. About 74% of familial conditions were covered. Overall, all constructive feedback has been adapted into the updated version.
Conclusion: Gia Su Suc Khoe has the potential to significantly improve healthcare delivery and outcomes in Vietnam.

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adult primary care; computer-based tool; family health history; genetic disorders; risk stratification

1. Introduction

The Human Genome Project has enabled a shift toward a preventative approach to diseases within primary care settings [1,2]. Human genomic profiling provides opportunities for diagnosing and treating complex chronic conditions [3]. However, focusing solely on complex conditions overlooks the broader implications of human genetics, which offer insights into both common and rare conditions [4]. Central to constructing a patient's genomic profile is their family health history (FHH). It serves as a tool for risk stratification [5–7] and helps identify appropriate tests, screenings, including genetic tests, based on presented symptoms [4].

Unfortunately, the traditional clinical approach for obtaining FHH heavily relies on primary care physicians [8–10], presenting several barriers. Inconsistency [11] and a lack of systematization [7,11] in collecting FHH have been documented, as it depends on various factors during patient consultations. Physicians often lack time to initiate FHH discussions or explore them in detail [9,10,12,13]. FHH discussions, when they occur, are typically one-off, leading to outdated information [9]. Moreover, physicians feel they have limited knowledge and skills to collect FHH and discuss disease risks comprehensively [2,8]. Some physicians perceive patients' understanding of their family history as a barrier to obtaining accurate FHH [12,14].

In light of the barriers highlighted in existing literature, there is a clear demand for a systematic and uniform method for recording FHH. Despite the growing use of genetic information in disease management and treatment in Vietnam, there is presently no established protocol for documenting FHH utilizing a pedigree format. To bridge this deficiency, we have devised and validated a tool named "Gia Su Suc Khoe" (GSSK), a self-administered and patient-centric computerized program for obtaining FHH. This paper delineates the conceptual framework, developmental journey, and validation process of GSSK, positioning it as the pioneering model for patient-entered family history documentation.

2. Materials & methods

2.1. Program design goals

GSSK represents our original work, distinct from any adaptations from elsewhere. Based on our clinical experience, insights from published literature, and analysis of

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existing programs, we have identified key characteristics essential for a FHH software program to be effective in clinical practice. These include streamlining clinical workflow by allowing patients to input their own FHH and integrating risk assessment for five common hereditary cancers [14,15]. Furthermore, by analyzing the insights and the limitations of published tools [16], we aimed to address and overcome the acknowledged limitations of these tools. With these considerations in mind, we have established specific design objectives for GSSK: (i) to create a user-friendly interface for collecting FHH data comprehensively, including a detailed three-generation pedigree with information such as age of disease onset, current age or age at death and cause of death for each relative, (ii) to offer risk assessment for prevalent genetic disorders.

Through these design goals, GSSK endeavors to empower families to efficiently gather valuable FHH information, potentially facilitating the collection of critical health data and assisting healthcare providers in making well-informed decisions and delivering personalized care.

2.2. GSSK tool description

GSSK, a stand-alone Web-based program, comprises two integral components: FHH collection and risk stratification for five common cancers. These components were developed in tandem to optimize their effectiveness.

The development of GSSK's FHH collection and decision support involved a multidisciplinary team comprising four genetic counselors with expertise in adult, pediatric, and cancer genetics, ten medical geneticists, and three information technology experts. This process facilitated consensus-building on which professional guidelines and expert opinions to base the algorithms, as well as determining which conditions to include.

Recommended by Rich and colleagues [4], the GSSK tool necessitates users to provide core, pertinent information, including: (1) compiling FHH spanning at least three generations, (2) age or year of birth, (3) age and cause of death, (4) relevant health data (such as height, weight, lifestyle habits and physical activities), (5) documented illnesses and age at diagnosis, (6) consanguinity and (7) pregnancy-related details.

2.3. FHH collection

The FHH collection component serves as the primary interface for patients. It employs a user-friendly web-based survey that begins by establishing the family's structure, capturing names and ages (current or age at death) for at least three generations of relatives. Subsequently, patients identify which relatives have been affected by any of the 51 predetermined conditions (Table 1). These conditions were meticulously selected by compiling a list of significant familial and hereditary conditions. To optimize the tool's effectiveness while minimizing user burden, only the top-ranked 51 conditions were integrated into GSSK (*Please refer to English content of GSSK in* Supplementary Table S2).

GSSK operates in full-screen mode to enhance user experience, presenting only questions and response fields without clutter from toolbars or menus. All fields are touch-screen enabled and fonts/buttons are designed to be large and easily readable. Additionally, the use of branching questionnaire logic allows GSSK to skip irrelevant survey question screens, reducing the time taken to complete the survey. Moreover, family health histories can be updated, and algorithms can be rerun as necessary, ensuring ongoing accuracy and relevance.

Moreover, the program provides well-established risk-stratified screening and preventive care strategies known for their significant clinical value. The qualitative risk assessment is formulated following broad guidelines, utilizing a comprehensive family history to stratify individuals for numerous preventable, prevalent genetic disorders (Table 2) [17]. Guided by these parameters, five pilot diseases – breast cancer, ovarian cancer, endometrial cancer, colon cancer, prostate cancer and the risk for hereditary cancer syndromes – were chosen to demonstrate the efficacy and acceptability of GSSK. The GSSK tool assesses the risk for 5 cancers, therefore, only the risk assessment for these will be reported in pop-up output while presented with no hidden results.

The decision support system delineates risk categories and corresponding action-oriented risk management strategies for five target cancers and hereditary cancer syndrome. These strategies are arranged in order of decreasing risk and include referral to genetic counseling, management of increased personal and familial risk by the provider, and routine population-based screening. An iterative algorithm was developed wherein patients meeting the criteria for genetic counseling referral are identified initially, followed by the identification of patients at familial or population risk. This approach ensures that individuals with the highest risk receive appropriate attention and intervention first, optimizing the allocation of resources and enhancing patient care.

2.4. Coding

The GSSK utilizes cutting-edge technology including Angular, .NET Core 8 and SQL Server 2022. Operating within a Windows Server 2022 environment with an IIS web server, our application offers a robust platform for administrators and clinical coordinators to securely

Table 1. Top-ranked 51 conditions integrated into Gia Su Suc Khoe.

	Conditions		
	Cardiovascular diseases		Cancers
1	Long QT Syndrome (LQTS)	27	Thyroid cancer
2	Dilated Cardiomyopathy (DCM)	28	Skin cancer
3	Hypertrophic Cardiomyopathy (HCM)	29	Brain cancer
4	Brugada Syndrome	30	Bone cancer
5	Atrial Fibrillation	31	Leukemia
6	Heart Arrhythmia	32	Multiple Myeloma
7	Myocardial infarction	33	Kidney cancer
8	Coronary Artery Disease	34	Liver cancer
9	Angina	35	Gastric cancer
10	Hypertension	36	Esophageal cancer
	Hematologic Diseases	37	Colorectal cancer
11	Beta Thalassemia	38	Prostate cancer
12	Alpha Thalassemia	39	Ovarian cancer
13	Hypercholesterolemia	40	Endometrial Cancer
14	Familial hypercholesterolemia	41	Breast cancer
15	Pulmonary Embolism	42	Lung cancer
16	Deep vein thrombosis		**Others**
17	Haemophilia	43	Irritable Bowel Syndrome (IBS)
	Neurological /Psychiatric diseases	44	Muscular Dystrophy
18	Psychosis	45	Osteoporosis
19	Convulsions	46	Maturity-Onset Diabetes of the Young (MODY)
20	Epilepsy	47	Diabetes Type 2
21	Autism Spectrum Disorder	48	Diabetes Type 1
22	Attention Deficit Hyperactivity Disorder (ADHD)	49	Crohn's Disease
23	Attention Deficit Disorder (ADD) without Hyperactivity	50	Chronic Obstructive Pulmonary Disease (COPD)
24	Parkinson's Disease	51	Asthma
25	Dementia		
26	Alzheimer's Disease		

Table 2. Guidelines of qualitative risk assessment in Gia Su Suc Khoe.

High risk	Moderate risk	Low risk
1. Premature disease[a] in a 1st degree relative. 2. Premature disease in a 2nd degree relative (coronary artery disease only). 3. Two affected 1st degree relatives. 4. A 1st degree relative with late/unknown onset of disease and an affected second degree relative with premature disease from the same lineage. 5. Two second degree maternal or paternal relatives with at least one having premature onset of disease. 6. Three or more affected maternal or paternal relatives. 7. The presence of a "moderate risk" family history on both sides of the pedigree.	1. A first degree relative with late or unknown disease onset. 2. Two second degree relatives from the same lineage with late or unknown disease onset.	1. No affected relatives. 2. Only one affected second degree relative from one or both sides of the pedigree. 3. No known family history. 4. Adopted individual with unknown family history.

[a]Premature disease: coronary artery disease onset $\leq$55 year in males, $\leq$65 year in females; stroke, noninsulin-dependent diabetes, colon and prostate cancer onset $\leq$50 year; breast, ovarian and endometrial cancer onset premenopausal or $\leq$50 year.
Pedigrees demonstrating clustering of different primary cancers consistent with a family cancer syndrome were high risk.
Pedigrees demonstrating clustering or cardiovascular diseases and noninsulin-dependent diabetes consistent with Syndrome X were considered high risk.
Data taken from [17].

access patient and questionnaire data. Key functionalities include updating patient contacts, such as letters and phone conversations, performing mail merges for introduction letters to potential participants, generating post-questionnaire pedigree and summary reports, and presenting vital patient tracking and quality evaluation metrics. These metrics encompass demographics, patient visit dates and completed questionnaires, as well as tracking patients who missed appointments or declined participation.

2.5. Initial user's survey & feedback

To enhance the collection of FHH and refine decision support algorithms and reports, pilot testing was conducted in multiple stages. Initially, the first phase entailed testing with three genetic counselors and two family-medicine doctors, followed by the second phase involving 20 community volunteers. Throughout this process, iterative direct feedback from providers and an online survey to gather feedback participants (Supplementary Table S1).

This valuable input was then utilized to optimize the content and design of the reports, ensuring they meet the needs and preferences of all stakeholders involved. Subsequently, a comprehensive 2-month pilot test was undertaken with 600 community volunteers during the third phase. The Institutional Review Board approved the study and volunteers were explained the implementation of GSSK and provided consent to join the survey before its start.

3. Results

During the validation and community pilot testing phases, three iterative cycles of feedback data were collected for GSSK revision.

Feedback from healthcare providers primarily centered around the quality, usability and accuracy of GSSK, particularly its risk-prediction algorithms and printed pedigrees. To evaluate the accuracy of the programming, coding, algorithms, and report outputs, each counselor inputted at least two sample cases into GSSK. They then reviewed the risk-prediction output and provided direct feedback to the study team via phone or mail. This process allowed for thorough assessment and refinement of the system to ensure its effectiveness and precision in clinical settings.

During the second phase of piloting, 20 individuals participated, including 15 females and 5 males, aged between 20 and 40 years old, and possessing at least a 12th-grade education. Using the online tool, they completed the FHH collection and provided feedback on its usage, design, and content via an online survey developed by the study team. This diverse group offered valuable insights into the tool's usability and effectiveness across various demographics. On average, participants took 10–15 minutes to complete GSSK. Impressively, 100% of respondents noted that the programming accurately reflected information from their family history and expressed satisfaction with the color and brightness. Additionally, 74% indicated that the surveyed diseases covered all aspects of their FHH. Moreover, over 50% expressed satisfaction with the overall usage, design, and question order.

Constructive feedback from volunteers included suggestions to increase font and button sizes, incorporate a "don't know" response option, emphasize important instructional words, enhance the visibility of the status bar, and simplify and organize GSSK's questions. In response to these suggestions, longer questions were broken into multiple shorter ones, questions about maternal and paternal relatives were organized more intuitively, and pop-up boxes were added to define diseases in lay terminology. Furthermore, the programming underwent revisions to enable users to easily remove mistakenly entered relatives and automatically save entered information, facilitating seamless navigation within the tool.

The final community pilot program received 574 family heath history collection through convenient sampling of volunteer white-collar employees. The implementation was uniformly positive, and minor changes were made for mismatching and designing. About 55 different diseases were recorded.

4. Discussion

Heath care providers are tasked with the systematic collection of FHH and the management of their patients' disease risks accordingly. However, numerous barriers at the provider, patient and system levels in primary care hinder the adoption of this seemingly straightforward yet complex activity [7–13]. This paper delineates the foundational objectives, evolution and systematic validation of GSSK, a computerized program enabling communities to input their FHH and offering risk assessment for common inherited cancers. GSSK addresses several barriers to obtaining high-quality family health histories and leveraging this information for risk assessment. Throughout its development, GSSK underwent iterative adaptations, including usability and comprehension testing with community volunteers, evaluation by genetic counselors, health care providers for usability, content and accuracy, and trials in clinical settings to assess feasibility, adoption and accuracy. The culmination of this process is a validated tool meticulously designed to facilitate the collection of FHH and the implementation of evidence-based prevention and screening guidelines in the fast-paced environment of clinical practices.

A structured FHH is crucial for precise risk assessment in individuals and their families. FHH serves as the cornerstone of precision medicine, as genetic and genomic testing should always be interpreted within the context of a patient's medical and family background. Increasingly, guidelines rely on risk stratification to inform prevention and screening strategies. The guideline for risk stratification was adopted from the work of Scheuner MT et al. [17], which indicated that assessing the risk of breast cancer, ovarian cancer, endometrial cancer, colon cancer and prostate cancer is feasible and potentially effective by considering personal family history. Overall, risk assessment provides significant benefits by raising awareness of risk associations, encouraging individuals to participate in general population screening, and offering the opportunity for earlier, more frequent, and intensive screening for diseases, particularly associated cancers. According to this guideline, GSSK also provides long-

term advantages through appropriate consultation. The guideline suggests that the presence of clustering of different primary cancers within one pedigree, indicative of a family cancer syndrome, should be classified as high risk. Additionally, even if only one cancer type occurs in the pedigree, the consultant can determine that the individual is at a higher risk for other related cancers, based on evidence of cancer syndromes. It is essential that GSSK results, along with the pedigree tree, are shared with a genetic counselor or family doctor. Armed with a well-curated health record, health care providers can pinpoint individuals with high risk of illness, offer tailored treatment recommendations, propose effective risk reduction measures and guide families in adopting lifestyle changes geared towards prevention.

The tool embodies four out of the seven characteristics delineated for the "ideal family history tool" by Rich and colleagues [4]. GSSK is crafted to be "patient-completed" and "adapted to patient age, gender, common conditions". It effectively "elicits specific patient concerns" and is engineered to be "Brief, understandable, easy to use", substantiated by our comprehensive pilot testing involving usability testing, genetic counselor review and community pilot. Crucially, the programming pedigree can be printed for dissemination among the user's family and/or healthcare provider for consultation, and the FHH can be saved and updated over time.

Several limitations of the study are noteworthy. Firstly, the majority of survey respondents had attained at least some college education, potentially limiting the generalizability of findings to broader demographics. Secondly, disparities in internet access and technological comfort were evident, particularly among older generations who may be less familiar with online platforms. Moreover, issues related to digital literacy were observed, as some participants struggled to effectively navigate online tools, especially when inputting intricate medical information. Future effectiveness trials should aim to recruit a more diverse demographic sample to comprehensively evaluate platform usage and its impact on enhancing community attitudes and engagement with FHH. It is worth mentioning that some expanded merits of previously designed tools were explored [16] including the features of shareable and editable across relatives and integration to personal medical records, which have not been facilitated in GSSK. However, the development of our tool is ongoing and refining. The incorporation of widespread surveys to understand user needs and the iterative process of adapting user experience and feedback are crucial steps. The potential integration of GSSK into the healthcare system indeed holds promise, benefiting both patients and the broader community.

For effective integration into clinical practice and population health management, the utilization of the GSSK tool requires endorsement and guidance from the Department of Health. This entails seamless integration with medical health records and adherence to pertinent regulations and guidelines to ensure the confidentiality of users' personal health data. In the next phase, we propose integrating the GSSK tool into medical health records. This integration into the healthcare system is essential to validate the tool's accuracy in implementing GSSK in the population.

5. Conclusion

Overall, the development and validation of a computer-based tool for obtaining FHH in Vietnam have the potential to significantly improve healthcare delivery and outcomes in the country. Ongoing development of GSSK must be paid to addressing clinical validity, clinical utility and privacy considerations to ensure its effectiveness and acceptance among the target population.

Article highlights

- Family health history (FHH) has been a primordial, non-expensive, and non-invasive risk stratification tool to inform prevention and screening strategies, especially for common genetic conditions.
- A new program developed in Vietnam namely Gia Su Suc Khoe (GSSK) not only utilizes a pedigree format to capture FHH but also performs risk assessments for various preventable and prevalent genetic disorders.
- GSSK is a self-administered, patient-centric and computerized program for obtaining Vietnamese FHH (https://giasusuckhoe.vn/), which receives positive feedback from genetic counselors and family-medicine doctors on its effectiveness, precision, and practical implications.
- GSSK becomes the pioneering program to help obtain FHH in Vietnam, which is believed to address several barriers, ensure high-quality information and leverage it for risk assessment.

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Writing disclosure

No writing assistance was utilized in the production of this manuscript.

Ethical conduct of research

Institutional ethic review boards from study hospitals have approved this work. Informed consent has been obtained from the participants.

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