Universität Basel

Research Project

The CASCADE cohort: a family-based cohort for investigating the use and impact of genetic testing, and the development of comprehensive interventions for hereditary breast/ovarian and Lynch syndromes in Switzerland

Third-party funded project

Project title The CASCADE cohort: a family-based cohort for investigating the use and impact of genetic testing, and the development of comprehensive interventions for hereditary breast/ovarian and Lynch syndromes in Switzerland
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Project Website www.swisscascade.ch
Project start 01.02.2018
Probable end 31.01.2021
Status Completed

Breast, colorectal, ovarian, and endometrial cancers constitute approximately 30% of newly diagnosed cancer cases in Switzerland, affecting more than 12,000 individuals annually. About 2-15% of incident cases are due to known pathogenic germline variants. Approximately 5% of breast cancer cases and 10-15% of epithelial ovarian cancer cases develop due to the BRCA genes, which are associated with Hereditary Breast and Ovarian Cancer (HBOC). Lynch syndrome (LS) accounts for about 2-5% of colorectal cancer and endometrial cancers, and increased risk for other cancers including ovarian and pancreatic. Germline pathogenic variants in the DNA Mismatch Repair (MMR) account for most LS cases. The availability of genetic services for HBOC and LS is a significant milestone for effective cancer prevention and control. Genetic counseling can educate patients and cancer-free individuals about cancer risk and management options according to mutation status. For every carrier of a pathogenic germline variant, first- and second-degree relatives and first cousins have increased probability of carrying the same mutation. Due to availability of genetic testing, blood relatives can be tested with 100% accuracy. Cancer predisposition cascade genetic screening is a sequential process of identifying and testing blood relatives of a known mutation carrier. Goals of cascade screening are to determine if untested relatives also carry the pathogenic variant and propose preventive and clinical management options to reduce morbidity and mortality, and to identify non-carrier relatives and exclude them from intensive medical interventions. This approach has been strongly supported by the Center for Disease Control and Prevention (CDC), Office for Public Health Genomics. In Switzerland less than 25% of cases indicative of HBOC or LS use genetic services, suggesting that many mutation carriers and their blood relatives may not benefit from advances in medical diagnostics. HBOC and LS patients can benefit from intensive surveillance, pharmacoprevention, and/or prophylactic surgery. Monitoring cancer surveillance of mutation carriers ensures adequate quantity and quality of cancer care and coordination of healthcare services. Moreover, penetrance of these pathogenic mutations varies, and is likely influenced by modifiable behavioral and psychosocial risk factors, such as smoking, alcohol, and unhelpful coping with cancerrelated stress (e.g., avoidance and withdrawal). However, information about practices related to cancer screening, risk reduction, and cancer surveillance, and about modifiable behavioral and psychosocial risk factors can be obtained only from prospective family-based studies. Currently, there are

no family-based cohorts for HBOC and LS in Switzerland. This critical information will provide evidence across the full continuum of cancer risk and support the development of targeted risk modification and preventive interventions. The purpose of this application is to support the development of the CASCADE cohort, a Swiss- and familybased cohort enriched for hereditary cancer risk by including members of families harboring germline pathogenic variants associated with HBOC and LS. The CASCADE cohort will facilitate the collection of epidemiological, cancer surveillance, behavioral, and psychosocial data, which over time will assist in finding sustainable solutions and developing interventions that optimize the Swiss healthcare system in preventing, managing and treating HBOC and LS. This application is submitted on behalf of the Swiss Cancer Predisposition Cascade Screening (CASCADE) Consortium, an inter-professional research consortium with scientists from basic, clinical, and social science from the German-, French-, and Italian-speaking regions of Switzerland. The purpose of the consortium is to investigate the use and impact of genetic testing for HBOC and LS in Switzerland, and develop and disseminate comprehensive interventions to families harboring these pathogenic variants. Members of the consortium are excellent researchers, clinicians, and scholars in the fields of HBOC and LS in Switzerland and internationally, and their pioneering studies support the necessity of cancer predisposition cascade genetic screening for HBOC and LS.

Financed by
Foundations and Associations

Follow-up project of 3472041 CASCADE - Cancer predisposition cascade genetic screening for Hereditary Breast and Ovarian Cancer and Lynch Syndrome in Switzerland

Add publication

Published results

4617589, Baroutsou, Vasiliki; Underhill-Blazey, Meghan L.; Appenzeller-Herzog, Christian; Katapodi, Maria C., Interventions Facilitating Family Communication of Genetic Testing Results and Cascade Screening in Hereditary Breast/Ovarian Cancer or Lynch Syndrome: A Systematic Review and Meta-Analysis, 2072-6694, Cancers, Publication: JournalArticle (Originalarbeit in einer wissenschaftlichen Zeitschrift)

4617622, Baroutsou, Vasiliki; Underhill-Blazey, Meghan L; Appenzeller-Herzog, Christian; Katapodi, Maria C, Interventions Facilitating Family Communication of Genetic Testing Results and Cascade Screening in Hereditary Breast/Ovarian Cancer or Lynch Syndrome: A Systematic Review and Meta-Analysis., 2072-6694, Cancers, Publication: JournalArticle (Originalarbeit in einer wissenschaftlichen Zeitschrift)

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