Implementation of public health genetic interventions

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Personal and Family History, and Genetic Testing - Tools for Disease Prevention and Control

- Personal health history
- Family health history
- Advances in genetic/genomic testing
- Pathogenic variants associated with hereditary syndromes
- Cascade genetic screening in blood relatives
- Disease prevention and control
Personal and Family History and Genetic Testing - Tools for Cancer Prevention and Control

About 2%-15% of breast, colorectal, endometrial, ovarian cancer cases are due to inherited syndromes

Approximately 1,800 new cases per year in Switzerland

- Very high probability for >1 cancer
- Early age onset <45 → consequences for life trajectory / finances
- Biological impact on blood relatives (FDR, SDR, First Cousins)
  (12.5% - 50% probability for inheriting the pathogenic variant)
Hereditary Breast/Ovarian Cancer - HBOC

Monogenic disorder - autosomal dominant germline mutations
Primarily **BRCA1, BRCA2, PALB2**

<table>
<thead>
<tr>
<th>Cancer Type</th>
<th>General Population Risk</th>
<th>Mutation Risk BRCA1</th>
<th>Mutation Risk BRCA2</th>
</tr>
</thead>
<tbody>
<tr>
<td>Breast</td>
<td>12%</td>
<td>50%-80%</td>
<td>40%-70%</td>
</tr>
<tr>
<td>Second primary breast</td>
<td>3.5% within 5 years Up to 11%</td>
<td>27% within 5 yrs</td>
<td>12% within 5 yrs 40%-50% at 20 yrs</td>
</tr>
<tr>
<td>Ovarian</td>
<td>1%-2%</td>
<td>24%-40%</td>
<td>11%-18%</td>
</tr>
<tr>
<td>Male breast</td>
<td>0.1%</td>
<td>1%-2%</td>
<td>5%-10%</td>
</tr>
<tr>
<td>Prostate</td>
<td>15% (N. European origin)</td>
<td>&lt;30%</td>
<td>&lt;39%</td>
</tr>
<tr>
<td></td>
<td>18% (African Americans)</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Pancreatic</td>
<td>0.50%</td>
<td>1%-3%</td>
<td>2%-7%</td>
</tr>
</tbody>
</table>

**Tumor suppressor genes**
Produce proteins that repair damaged DNA. Mutations in these genes lead to the accumulation of genetic defects that allow cells to grow and divide uncontrollably.
Lynch Syndrome

Monogenic disorder – autosomal dominant germline mutations in DNA mismatch repair (MMR) genes:
- **MLH1** (MutL homolog 1), Chromosome 3p21
- **MSH2** (MutS homolog 2), Chromosome 2p16
- **MSH6** (MutS homolog 6), Chromosome 2p16
- **PMS2** (postmeiotic segregation 2), Chromosome 7p22

Up to 90% in 30 patients with colorectal cancer has Lynch Syndrome
Recommendations for Genetic Screening: US Preventive Services Task Force and Centers for Disease Control and Prevention

Tier 1 Cancer Genetic Syndromes HBOC, LS

positive impact on public health - evidence-based guidelines
Easily detectable, acceptable, actionable

➢ Systematic screen personal and family history for HBOC, LS
➢ If positive, genetic counseling and genetic testing
➢ If testing positive, counseling for risk management
➢ Systematic cascade genetic screening of asymptomatic at-risk blood relatives

NELSON ET AL. ANN INTERN MED 2013; KHOURY MJ, EVANS JP. JAMA 2015
Identify individuals carrying a germline pathogenic variant associated with HBOC or LS

Extend genetic testing to his/her asymptomatic blood relatives

Offer risk management options to positive cases and exclude true negatives from increased surveillance

Due to privacy laws, communication of genetic test results to at-risk relatives can be ONLY through the mutation carrier

Barriers to Cancer Predisposition
Cascade Genetic Screening

- SES
- Decision making
- Screening/disease management
- Family support and communication

- Lack of genetic/genomic education
- Clinical management skills

- Availability, accessibility, acceptability
- Coordination of services
- Continuation of care
- Legislation, HTA
- Public health awareness

Individual - Family
Healthcare providers
Healthcare – Insurance system

Micro – Meso- Macro- level
Barriers to Cancer Predisposition
Cascade Genetic Screening

<table>
<thead>
<tr>
<th>Individual - Family</th>
<th>Healthcare providers</th>
<th>Healthcare – Insurance system</th>
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<tr>
<td>• SES</td>
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<tr>
<td>• Family support and communication</td>
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<tr>
<td>• Public health awareness</td>
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</table>

Micro – Meso- Macro- level
Screening and Genetic Testing in Young Breast Cancer Survivors and Relatives

Funding: Centers for Disease Control and Prevention (PI: Katapodi, 2011)

University of Michigan and Michigan Cancer Genomics Program

Community outreach to increase genetic testing and cancer surveillance in women with breast cancer < 45 y.o. and blood relatives

- Recruitment from Michigan cancer registry
- Random sample female breast cancer < 45 y.o.
- Purposeful sample 1-2 relatives (FDR or SDR)/ patient
- Randomized unit: Family
- Targeted (generic) vs. Tailored (person-specific) messages
Cancer Genetic Services in Switzerland

- 11% of all Swiss breast cancer patients have genetic testing
- 25% of breast cancer patients with a strong family history
- Lower numbers for Lynch syndrome
 CASCADE Consortium (est. 2016) - working association researchers, clinicians, community professionals, educators, students

**Goals** are to:

- Support research related to cancer predisposition genetic screening and care continuum
- Foster collaboration among health - community professionals
- Disseminate scientific advancements - scientists, practitioners, patients, families, healthcare institutions, and involved stakeholders
- Foster the development of researchers and clinicians through mentorship, access to data, and collaborative studies
Aim 1: Family-based cohort of HBOC and LS mutation carriers and at-risk relatives

Aim 2: Interventions for access to genetic services and cascade testing

Aim 3: Interventions for behavioral - psychosocial outcomes, quality of life

Cancer Predisposition Cascade Genetic Screening in Switzerland Hereditary Breast/Ovarian Cancer & Lynch Syndromes
CANCER PREDISPOSITION CASCADE GENETIC SCREENING IN SWITZERLAND HEREDITARY BREAST/OVARIAN CANCER & LYNCH SYNDROMES

Aim 1: Family-based cohort of HBOC and LS mutation carriers and at-risk relatives

(confirmed mutation carriers, untested relatives, true negatives)

Benefits of family-based cohort:

- enriched for hereditary cancer risk
- captures risk associated with family history in distant relatives and age of cancer onset
- study gene-environment interactions at heterogeneous levels of risk
- behavioral and psychosocial outcomes
- practices related to cancer screening and risk reduction
- facilitate translation of research findings into clinical practice
Flow of Assessments

1. Identification of Mutation Carriers from Genetic Testing Clinics
2. Invitation Letter signed and sent from Physician – Medical Director
3. K-CASCADE Headquarters – Informed Consent or Refusal Form
4. Informed Consent
   - Reasons for Refusal
   - NO
   - YES
   - I would like to complete a survey asking some of my personal information
   - K-CASCADE (please, check the box with your preference)
   - online, in a secure website
   - in paper and pencil format
   - Deutsch · Français · Italiano · English
   - I would like to invite some of my blood relatives to the study
   - I would like to take part with my relatives in a focus group and preview a web-based program (Family Gene Toolkit®), if selected by the research team
   - I would like to participate in the CASCADE Cohort by completing a similar survey once a year for five years
   - I would like to receive an annual newsletter with progress and findings of the study

5. You can reach me in the following ways:
   - Name: ____________________________

6. K-CASCADE Baseline Survey

Follow-up
CANCER PREDISPOSITION CASCADE GENETIC SCREENING IN SWITZERLAND HEREDITARY BREAST/OVARIAN CANCER & LYNCH SYNDROMES

Aim 1: Family-based cohort of HBOC and LS mutation carriers and at-risk relatives

Surveys

- Demographics
- Clinical characteristics
- Cancer status
- Surveillance
- Access - barriers to services
- Decision making
- Family engagement
- Quality of life

Focus groups - Interviews

- Perceptions about providers’ role and communication of genetic cancer risk
- Disclosure of genetic risk to at-risk relatives
- German, French, Italian
### Characteristics of Participants (April 2020)

<table>
<thead>
<tr>
<th></th>
<th>HBOC n=243</th>
<th>Lynch Syndrome n=50</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Female</strong></td>
<td>202 (83%)</td>
<td>30 (60%)</td>
</tr>
<tr>
<td><strong>Caucasian</strong></td>
<td>200 (82%)</td>
<td>40 (80%)</td>
</tr>
<tr>
<td><strong>Cancer Diagnosis</strong></td>
<td>121 (50%)</td>
<td>38 (76%)</td>
</tr>
<tr>
<td><strong>Breast</strong></td>
<td>77 (64%)</td>
<td>2 (5%)</td>
</tr>
<tr>
<td><strong>Ovarian</strong></td>
<td>26 (22%)</td>
<td>4 (11%)</td>
</tr>
<tr>
<td><strong>Pancreatic</strong></td>
<td>2 (2%)</td>
<td>1 (2%)</td>
</tr>
<tr>
<td><strong>Colorectal</strong></td>
<td>2 (2%)</td>
<td>24 (63%)</td>
</tr>
<tr>
<td><strong>Had genetic testing</strong></td>
<td>206 (85%)</td>
<td>49 (98%)</td>
</tr>
<tr>
<td><strong>Pathogenic variant</strong></td>
<td>189 (92%)</td>
<td>44 (90%)</td>
</tr>
<tr>
<td><strong>FDR (survey data)</strong></td>
<td>1238</td>
<td></td>
</tr>
<tr>
<td><strong>Willing to invite</strong></td>
<td>767 (62%)</td>
<td></td>
</tr>
<tr>
<td><strong>Willing to invite and eligible</strong></td>
<td>702 (57%)</td>
<td></td>
</tr>
<tr>
<td><strong>Accept participation</strong></td>
<td>351 (46%)</td>
<td></td>
</tr>
</tbody>
</table>
First Findings (April 2020)

- 65% of mutation carriers shared test results with some relatives
- 40% of mutation carriers do no remember receiving a recommendation for cascade genetic testing of relatives
- Providers address communication to relatives in a quick and non-detailed way; lack of continuity
- Family communication is complex and selective. It is subject to certain logics (e.g. “protection”) that overshadow the responsibility to communicate
- Females and those with greater genetic literacy are more likely to discuss with closer relatives
- In case of illness, the weight given to family communication is relative due to other concerns and priorities related to own health or to health of closest family members

Interviews n=20
Cascade Genetic Testing

Aim 2: Interventions for access to genetic services and cascade testing

Access to services is a multifactorial problem – Micro- Meso- Macro- Levels

- SES
- Decision making
- Screening/ disease management
- Family support and communication

Due to privacy laws, communication of genetic test results to at-risk relatives can be ONLY through the mutation carrier
The DIALOGUE Study: Using digital health to improve care for families with predisposition to hereditary cancer

SNSF-NRF Innovation Funding Program – Swiss-Korean Bilateral Collaboration

Web-based platform based on the Family Gene Toolkit

M 1: Knowledge of cancer genetics
M 2: Decisional support for genetic testing
M 3: Active coping with challenges
M 4: Skills-building communication training
M 5: Cancer risk management

Dose, duration: self-paced, within 4 weeks

Device agnostic (accessible via PC, mobile, tablet etc.)

Active comparator: www.kintalk.org
Development of a Web-based Family Intervention for BRCA Carriers and Their Biological Relatives: Acceptability, Feasibility, and Usability Study

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Abstract

Background: Carriers of breast cancer gene (BRCa) mutations are asked to communicate genetic test results to their biological relatives to increase awareness of cancer risk and promote use of genetic services. This process is highly variable from family to family. Interventions that support communication of genetic test results, coping, and offer decision support in families harboring a pathogenic variant may contribute to effective management of hereditary cancer.
The DIALOGUE Study: Adaptation of the Family Gene Toolkit

Focus groups basis for adaptation and tailoring of Family Gene Toolkit
Expert clinicians n = 6-10
HBOC carriers (n=10 -12) and at-risk relatives (n=10 -12)

Usability testing (n = 5-6): “Think aloud” method

Acceptability testing (n= 5-10): Clarity, appropriate length, level of detail, relevance, interest, satisfaction
1-7 Likert scale
The DIALOGUE Study: Message Tailoring

Targeted (generic) messages

• Limited variability in predictors

Tailored (person-specific) messages

• Variability in predictors
  • Identify predictors based on theory of stress and coping and family adaptation in genetic illness
  • Select predictors for tailoring based on variability of responses from focus groups and surveys

<table>
<thead>
<tr>
<th>Strategy</th>
<th>Sample message (tailored elements in <strong>bold</strong>)</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Personalization</strong></td>
<td></td>
</tr>
<tr>
<td>Cancer type</td>
<td>When someone has <strong>ovarian cancer</strong>, it affects the whole family, especially her <strong>underage children</strong></td>
</tr>
<tr>
<td>Mutation</td>
<td><strong>BRCA2</strong> mutations can be passed on to <strong>sons</strong> and <strong>daughters</strong></td>
</tr>
<tr>
<td><strong>Feedback</strong></td>
<td></td>
</tr>
<tr>
<td>Active coping</td>
<td>When you face difficult situations, you often <strong>try to find more information</strong></td>
</tr>
<tr>
<td>Passive coping</td>
<td>When you face difficult situations, you often like to <strong>withdraw and not discuss about the problem</strong></td>
</tr>
</tbody>
</table>
https://swisscascade.ch or https://k-cascade.kr

Four languages DE, FR, IT, EN
Korean to be added
The DIALOGUE Study: Cluster RCT for efficacy of adapted DIALOGUE platform
Randomization at the family level

Sample
Mutation carriers n=114 (expected 4 females : 1 male)
Cancer-free or have cancer
(expected 5 breast : 1 ovarian cancer)

Excluded
No at-risk relatives, no Husbands / partners
Mental illness
No access to the internet

DIALOGUE platform
Active comparator
The DIALOGUE Study:
Primary and secondary outcomes at 2 and 6 months

<table>
<thead>
<tr>
<th>Concepts</th>
<th>Instruments</th>
<th>Cronbach's alpha</th>
<th>Test- Retest Reliability</th>
<th>Assessment</th>
</tr>
</thead>
<tbody>
<tr>
<td>PRIMARY OUTCOMES</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Psychological distress</td>
<td>Profile of Mood States (POMS-SF) (103) 37 items, 7-point Likert scale</td>
<td>0.82-0.91</td>
<td>-</td>
<td>✓</td>
</tr>
<tr>
<td>Proportion of informed relatives</td>
<td>Self-Report</td>
<td>N/A</td>
<td>N/A</td>
<td>✓</td>
</tr>
<tr>
<td>Intention to inform relatives</td>
<td>Informing Relatives Inventory (102) 68 items, 7-point Likert scale</td>
<td>0.82-0.92</td>
<td>-</td>
<td>✓</td>
</tr>
<tr>
<td>Intention to have genetic testing (applicable for untested relatives)</td>
<td>1 item, 7-point Likert scale</td>
<td>N/A</td>
<td>N/A</td>
<td>✓</td>
</tr>
<tr>
<td>SECONDARY OUTCOMES</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Knowledge of breast cancer risk factors and genetics</td>
<td>Risk Factor Knowledge Index (38) 17 items, True, False, Don’t Know</td>
<td>0.89</td>
<td>0.85</td>
<td>✓</td>
</tr>
<tr>
<td>Breast Cancer Genetics Index (104) 12 items, True, False, Don’t Know</td>
<td>0.82</td>
<td>0.81</td>
<td>✓</td>
<td>✓</td>
</tr>
<tr>
<td>Coping with stressful events</td>
<td>Brief Cope (105) 25 items, 7-point Likert scale</td>
<td>0.71-0.90</td>
<td>0.71-0.85</td>
<td>✓</td>
</tr>
<tr>
<td>Decision making</td>
<td>Decisional Conflict Scale – Genetic Testing (106) (for untested individuals) 16 items, 7-point Likert scale</td>
<td>0.96</td>
<td>-</td>
<td>✓</td>
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<tr>
<td></td>
<td>Decisional Regret – Genetic Testing (107) (for individuals that had genetic testing) 5 items, 7-point Likert scale</td>
<td>0.87</td>
<td>-</td>
<td>✓</td>
</tr>
<tr>
<td>Quality of Life</td>
<td>SF-12 summary score (subdomains will be assessed purely exploratory) (108) 12 items, multiple point Likert scale</td>
<td>0.83</td>
<td>-</td>
<td>✓</td>
</tr>
<tr>
<td>INTERVENTION EVALUATION</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Evaluation of intervention acceptability</td>
<td>Intervention acceptability, interest, usefulness, level of detail, relevance, and satisfaction (92) 15 items, 7-point Likert scale</td>
<td>-</td>
<td>-</td>
<td>✓</td>
</tr>
</tbody>
</table>
## The DIALOGUE Study:
Theory-based tailoring variables

<table>
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<tr>
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<tr>
<td><strong>DEMOGRAPHICS</strong></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Demographics, Personal, Family Cancer History</td>
<td>Self-Report (114)</td>
<td></td>
<td></td>
<td>✓</td>
</tr>
<tr>
<td><strong>TAILORING VARIABLES</strong></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Type of relationship - proband and relative(s)</td>
<td>Self-Report</td>
<td>N/A</td>
<td>N/A</td>
<td>✓</td>
</tr>
<tr>
<td>Perceived risk</td>
<td>Perceived Cancer Risk (69)</td>
<td>N/A</td>
<td>N/A</td>
<td>✓</td>
</tr>
<tr>
<td>Fear of cancer recurrence (for cancer patients)</td>
<td>Concerns About Recurrence Scale (CARS) (109)</td>
<td>0.93</td>
<td>0.91</td>
<td>✓</td>
</tr>
<tr>
<td>Self-efficacy dealing with cancer (for cancer patients)</td>
<td>Self-Efficacy – Breast Cancer (110)</td>
<td>0.80</td>
<td>0.71</td>
<td>✓</td>
</tr>
<tr>
<td>Self-efficacy using genetic services (for cancer patients)</td>
<td>Self-efficacy using genetic services</td>
<td>N/A</td>
<td>N/A</td>
<td></td>
</tr>
<tr>
<td>Family support</td>
<td>Family Support in Illness (111)</td>
<td>0.86</td>
<td>0.83</td>
<td>✓</td>
</tr>
<tr>
<td>Family hardness</td>
<td>Family Hardiness Index (112)</td>
<td>0.90</td>
<td>0.78 - 0.86</td>
<td>✓</td>
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<tr>
<td>Satisfaction with genetic counseling (for tested individuals)</td>
<td>Multidimensional Impact of Cancer Risk Assessment (MICRA) (113)</td>
<td>0.75 – 0.86</td>
<td>-</td>
<td>✓</td>
</tr>
<tr>
<td>Barriers and facilitators for genetic services</td>
<td>Barriers and facilitators for genetic services (37)</td>
<td>N/A</td>
<td>N/A</td>
<td></td>
</tr>
</tbody>
</table>
Flow of Assessments

Informed Consent

Reasons for Refusal

NO YES
- I would like to complete a survey asking some of my personal information
  - K-CASCADE (please, check the box with your preference)
  - online, in a secure website
  - in paper and pencil format
  - Deutsch
  - Français
  - Italiano
  - English
- I would like to invite some of my blood relatives to the study
- I would like to take part with my relatives in a focus group and preview a web-based program (Family Gene Toolkit®), if selected by the research team
- I would like to participate in the CASCADE Cohort by completing a similar survey once a year for five years
- I would like to receive an annual newsletter with progress and findings of the study

You can reach me in the following ways:
Name: __________________________

K-CASCADE Baseline Survey

Follow-up

DIALOGUE study
Randomization to adapted Family Gene Toolkit or adapted KinTalk

Adapted Family Gene Toolkit
- Content: 5 Modules (tailored)
- Dose: variable (recorded)
- Duration: 4 weeks
- Exit Survey – Invites Relatives

Adapted KinTalk (comparator)
- Content: Static Website
- Dose: variable (recorded)
- Duration: 4 weeks
- Exit Survey – Invites Relatives
The DIALOGUE Study:
Implementation and dissemination of DIALOGUE platform
RE-AIM Framework [www.re-aim.org](http://www.re-aim.org)
The DIALOGUE Study:  
Implementation and dissemination of DIALOGUE platform

RE-AIM Framework [www.re-aim.org](http://www.re-aim.org)

<table>
<thead>
<tr>
<th>RE-AIM dimension</th>
<th>Definition</th>
<th>Outcomes to be measured throughout the study</th>
<th>Strategies to be implemented to enhance future dissemination and implementation</th>
</tr>
</thead>
</table>
| **Reach** (individual level) | The absolute number, proportion, and representativeness of individuals willing to participate | • Response rate of mutation carriers  
• Number of relatives accessing the website(s)  
• Demographic, linguistic characteristics, region  
• Response rate to K-CASCADE | • Assess reasons for refusals (refusal form)  
• Mini-interview with those who decline participation  
• Help individuals set up free email accounts (Gmail etc.)  
• Post study advertisers to clinical settings |
| **Effectiveness** (individual level) | The impact of the intervention on outcomes, including negative effects, quality of life, economic outcomes, subgroup effects | • Assess times participants accessed each module  
• Assess number of “relative invites” initiated through the website  
• Evaluate acceptability, interest, usefulness, level of detail, relevance, and satisfaction at the follow up survey  
• Evaluate for potentially negative outcomes in the follow up survey (open-ended question) | • Individual tailoring and linguistic tailoring  
• Ongoing technical support to participants  
• Optimal maintenance of the online platform without interruptions |
| **Adoption** (setting, staff, or organization level) | The absolute number, proportion, and representativeness of settings and intervention agents who are willing to participate | • Number of clinicians and clinical settings willing to participate in the study  
• Diversity (geographic, linguistic, etc.) in participating settings | • Develop recruitment materials for clinical settings outlining the FGT benefits and K-CASCADE  
• Advertise the program within the SAKK network for Switzerland and the KOHBRA network for Korea  
• Conduct mini-interviews with participating and non-participating clinical settings and assess the need for further customization |
| **Implementation** (setting, staff, or organization level) | The intervention agents “fidelity” to the key elements of an intervention. This includes consistency of delivery as intended, adaptations made, and the time and cost of the intervention. | • Monitor referrals of mutation carriers from different clinical sites  
• Evaluate the cost for adapting modules for other hereditary cancer syndromes e.g., Lynch syndrome | • Provide demonstrations of the program to clinical settings |
| **Maintenance** (individual and setting levels) | | • Assess resources needed to maintain the website  
• Assess number of visits per month/year | • Incorporate HBOC support groups in each country  
• Seek feedback from clinical settings about rates of cascade genetic testing |

- Assess quality of life for calculating QALYs in future cost-effectiveness analysis
- Number of clinicians and clinical settings willing to participate in the study
- Diversity (geographic, linguistic, etc.) in participating settings
- Develop recruitment materials for clinical settings outlining the FGT benefits and K-CASCADE
- Advertise the program within the SAKK network for Switzerland and the KOHBRA network for Korea
- Conduct mini-interviews with participating and non-participating clinical settings and assess the need for further customization
- Provide demonstrations of the program to clinical settings
- Incorporate HBOC support groups in each country
- Seek feedback from clinical settings about rates of cascade genetic testing
Access to Cascade Genetic Screening in Switzerland

- Biological impact on blood relatives (FDR, SDR, TDR)
  (12.5% - 50% probability for inheriting the pathogenic variant)
- Cost of full sequence genetic testing ~ 3,500 CHF
- Cost of targeted genetic testing ~ 450 CHF
- No insurance coverage for SDR and TDR (50% missed relatives)

- Availability, accessibility, acceptability
- Coordination of services
- Continuation of care
- Legislation, HTA
- Public health awareness

Healthcare – Insurance system

SSPH+ ETH Zürich
Access to Cascade Genetic Screening in Switzerland

- Two studies currently examine the cost-effectiveness of cascade genetic testing for HBOC and LS in Switzerland.

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Evaluation of Genomic Applications in Practice and Prevention (EGAPP): Implementation and Evaluation of a Model Approach

The EGAPP initiative was launched by the CDC Office of Public Health Genomics in the fall of 2004. The initiative’s goal is to establish and evaluate a systematic, evidence-based process for assessing genetic tests and other applications of genomic technology in transition from research to clinical and public health practice. EGAPP also aims to integrate:

- existing recommendations on implementation of genetic tests from professional organizations and advisory committees,¹²³⁴
- knowledge and experience gained from existing processes for evaluation and appraisal (e.g., US Preventive Services Task Force, CDC’s Task Force on Community Preventive Services), previous CDC initiatives (e.g., the ACCE process for assembling and analyzing data on genetic tests), and the international health technology assessment experience.

Why is genetic testing a public health issue?
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