ACCESS:

A practice-guiding framework for overcoming disparities in genomic healthcare

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On behalf of the International Nursing CASCADE Consortium

Global Genomics Nursing Alliance – G2NA
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Conflict of Interest and Funding

No Conflict of Interest to Declare

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The genomic era

- Understand health and illness
- Improve diagnosis
- Tailor treatments
- Improve outcomes

- Large scale genomic research programs
- Big data and computational science methodologies
- Government-academic collaborations

Fu et al. 2020 Int J Nurs Sciences; NHGRI 2020

Access to genomic services

- In 2015 ~ 4.8 million Americans have used cancer-related genetic services
  - 2.1 million (97% women) for HBOC
  - 1.3 million (51% women) for colorectal cancer (Lynch syndrome)
  - 1.4 million (40% women) for other cancers
  - Disparities - sex, race/ethnicity, insurance, citizenship, education, age, cancer status

- Reasons for requesting genetic services
  - 66% healthcare provider/ clinician recommendation
  - 12% self-referral
  - 10% family request
  - 5% media influences

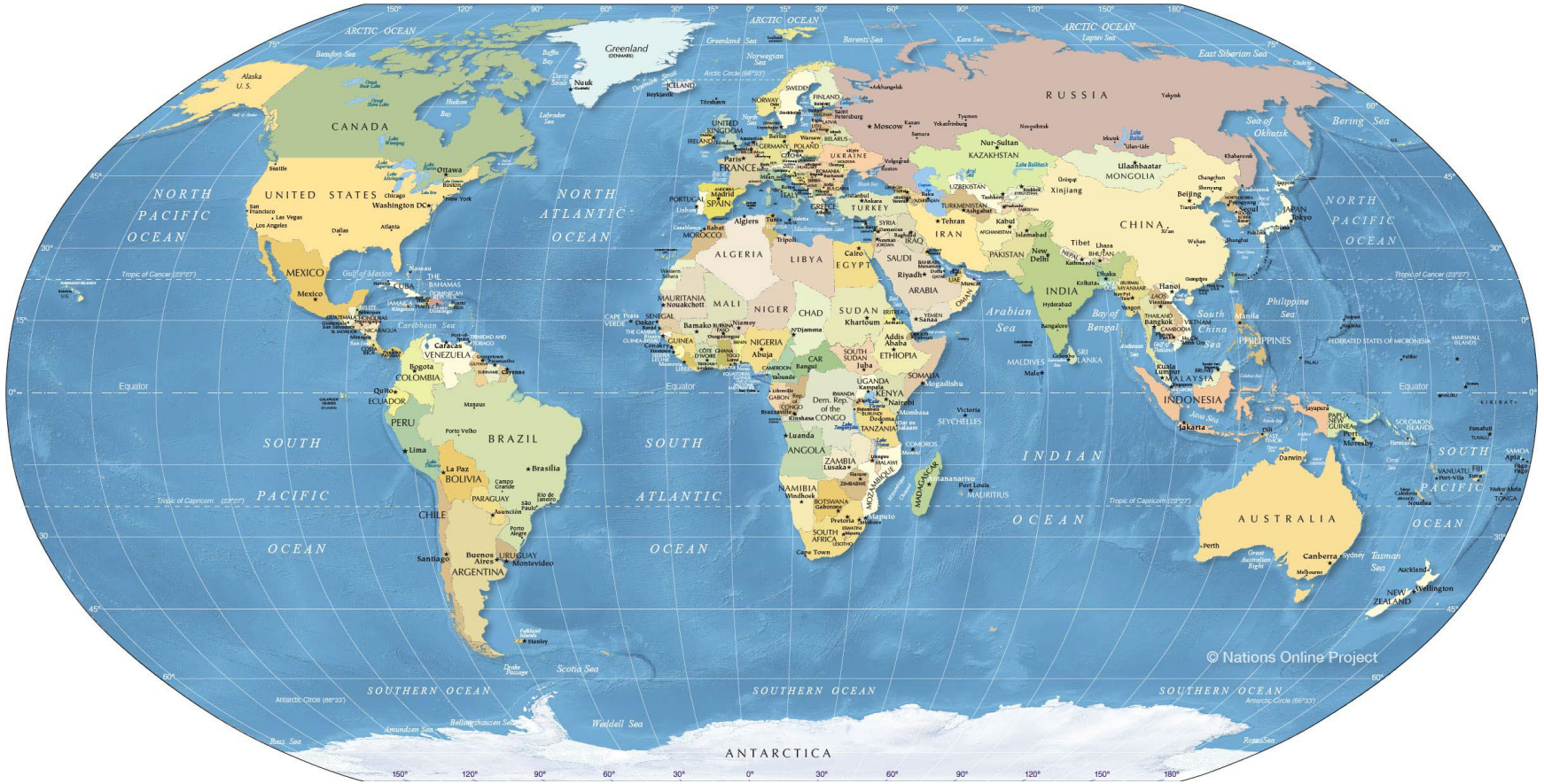
Stamp et al. Cancer Med 2019; Hinchcliff et al. Gynecol Oncol 2019

Increasing genomic healthcare disparities

- Growing disparities in accessing genomic care for **minority populations**
- Inadequate **diversity** in represented populations in genomic research and healthcare workforce
- Lack of genomic **services and infrastructure** outside traditional research centers and in diverse healthcare settings and underserved communities
- Inadequate **engagement** of communities and providers

Health Systems and Provider-related	Economic and Finances
Lack of … • ….capacity to care for expanding population • ….awareness and competencies • ….genetics specialists • ….appropriate referrals • ….effective patient-provider communication	• High out-of-pocket expenses • Inadequate insurance coverage • Insurance discrimination (e.g. life and disability insurance)
Societal	**Individual and Interpersonal**
• Language barriers • Distrust of healthcare providers • Discrimination and stigma • Privacy and confidentiality concerns	• Communication of family risk • Distress related to genetic testing (e.g. decisional conflict) • Family concerns (e.g. conflict, at-risk relatives) • Low genetic literacy and numeracy

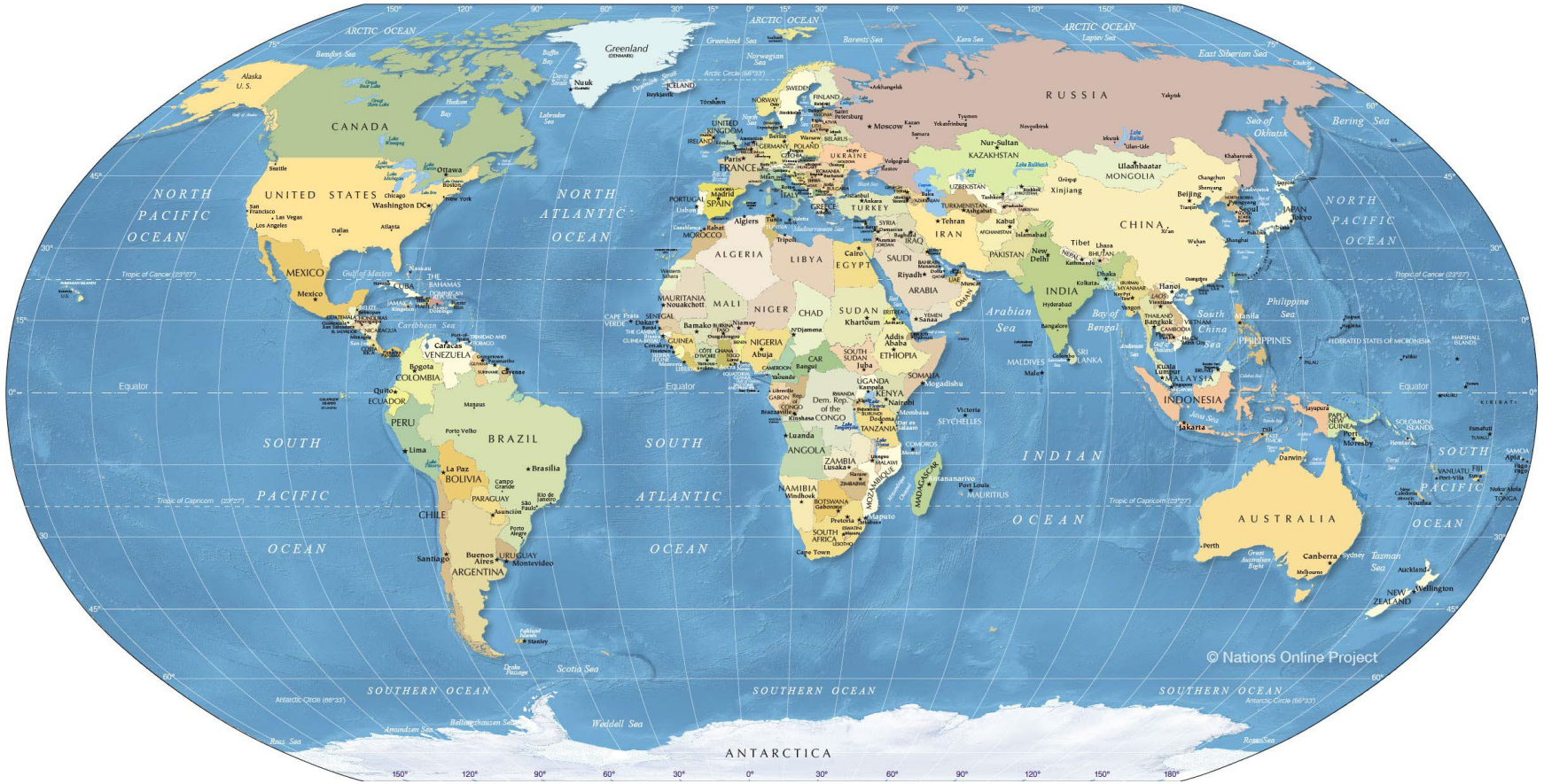
NASEM 2018; Jooma et al. 2019 Ethn Disease



How do we ensure that people around the world

benefit from advances in genomic technologies?

How do we ensure **access** to specialized genomic services?



Nurses are the most numerous healthcare providers globally

Nurse are the most trusted healthcare professionals in the US

Diverse professional roles and settings

Genomics across the lifespan and into routine (nursing) practice

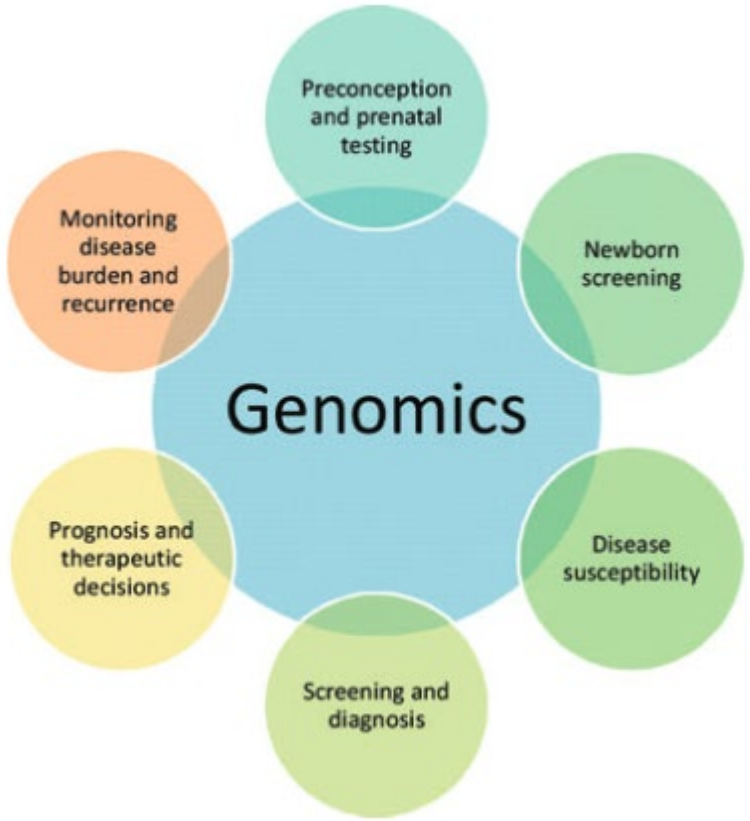


Figure. Areas of integration of genomic information in clinical practice.

Calzone et al. 2013 J Nurs Scholal; Tonkin et al. 2020 J Nurs Scholar; Kurnat-Thoma et al. 2021; Nurs Outlook

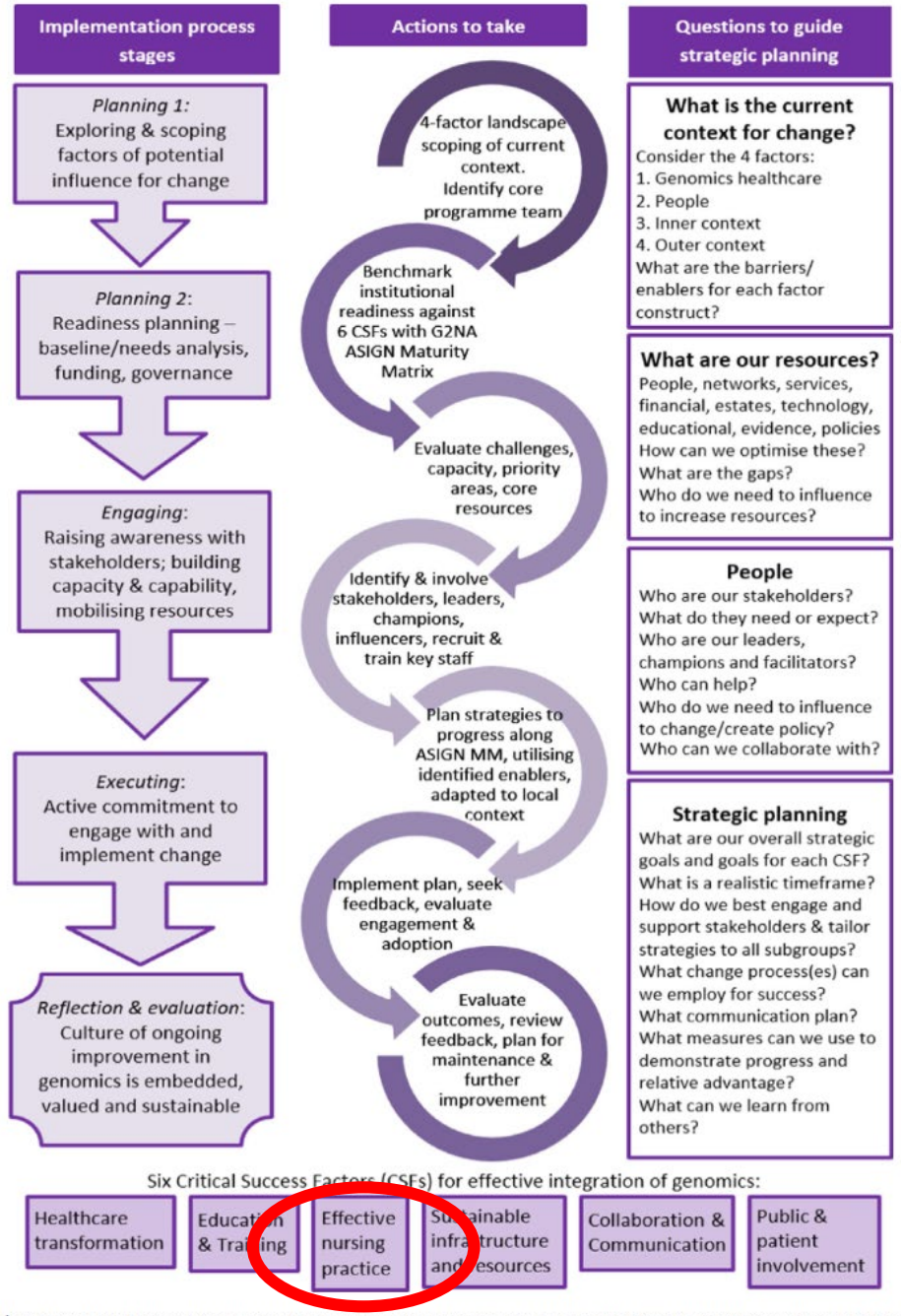


Figure 1. Global Genomics Nursing Alliance Roadmap for nurse leaders to implement genomics across nursing. [Colour figure can be viewed at wileyonlinelibrary.com]

International Nursing CASCADE Consortium

International nursing collaboration to accelerate genomics integration in practice

Organically formed from 2012 onward – cascade of collaborations

Common areas of research

- influence of genomic technologies on medical decision-making of underserved individuals, e.g., rare diseases, ethnic and racial minorities
- expanding genomic healthcare to families and implications for family communication and cascade screening of relatives
- implementing genomic healthcare across diverse cultures and organizational structures e.g., fee-for-service, public health genetic screening programs

International Nursing CASCADE Consortium

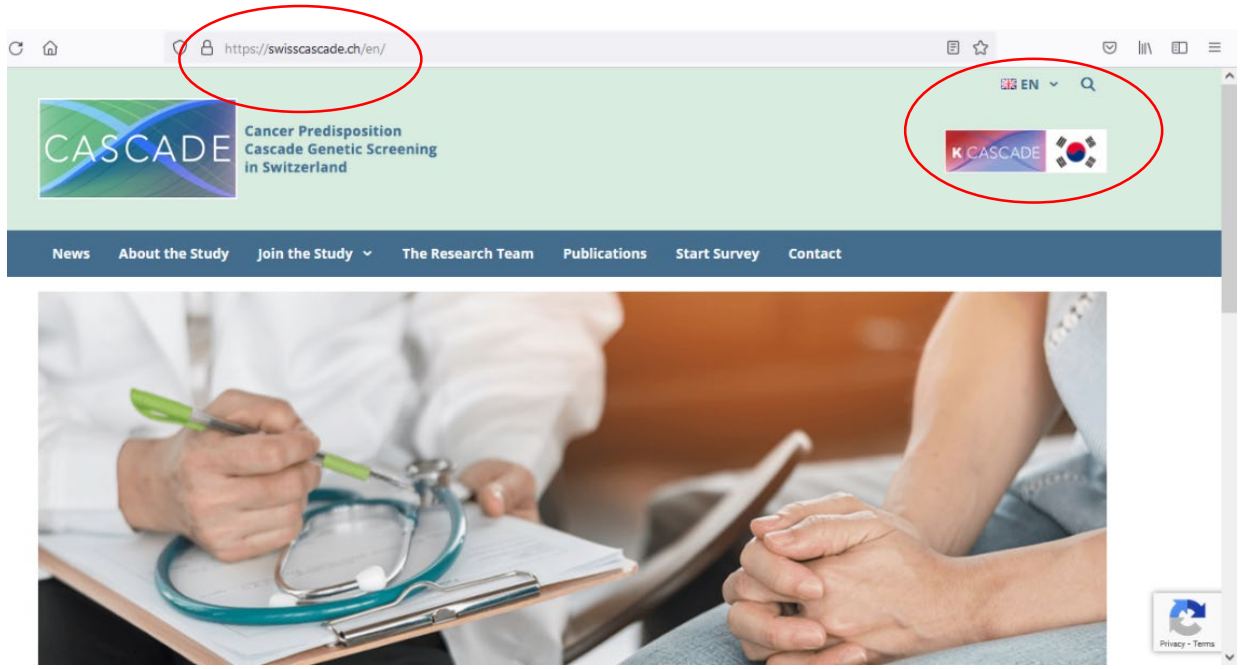
Diverse academic and clinical settings i.e., Israel, Korea, Switzerland, and U.S.

≥ 18 years of scholarship focused on genetics and genomics

Collectively ≥ 60 papers, mostly data-based

Diverse clinical populations with “common” and “rare” genetic and genomic disorders across the lifespan that are life-threatening or life-altering

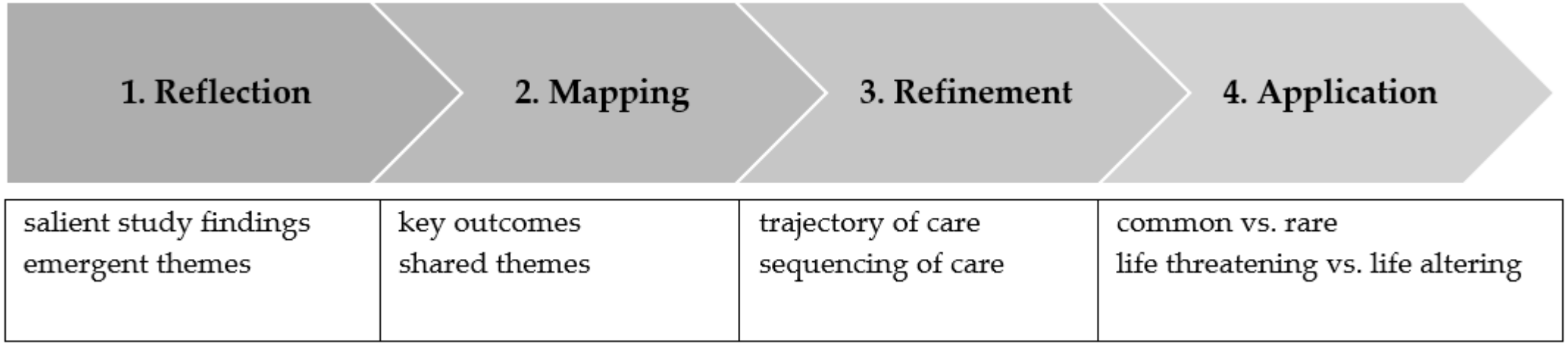
- HBOC, Lynch syndrome
- Cystic fibrosis
- Parkinson’s disease
- Kallmann syndrome
- Tay Sachs
- Canavan disease
- Fragile X-syndrome
- Spinal Muscular Atrophy



Development of the ACCESS framework

Over the course of 2+ years

- Broad themes running through studies – outcomes related to genomic disparities
- Themes examined for similarities across countries – healthcare systems and cultures
- Themes were organized according to the continuum of care – prevention, treatment, survivorship
- Framework was tested with healthy adults given hypothetical genetic testing scenarios – “common : rare” disease and “life-threatening : life-altering” paradigms



Outcomes related to genomic disparities and nursing care

Outcome	Nursing care element	Examples
Access to services	Identification of at-risk individuals	3 generation pedigree
	Genetic literacy and decisional support	Red flags, rule of "too - two"
	Referrals, connect primary care to specialized services	Underrepresented populations
	Advocacy for reimbursement, insurance coverage	Genomic literacy, lay language
		Informed decision making
		Teach-back strategies
		Digital technologies
		Advocacy for reimbursement and coverage
		National insurance coverage

Swiss CASCADE and K-CASCADE Cohort

HBOC and Lynch syndrome cases: who recommended that you have genetic testing?

Tested individuals:

83% Index cases

17% Relatives

- (n=)
- (n=)

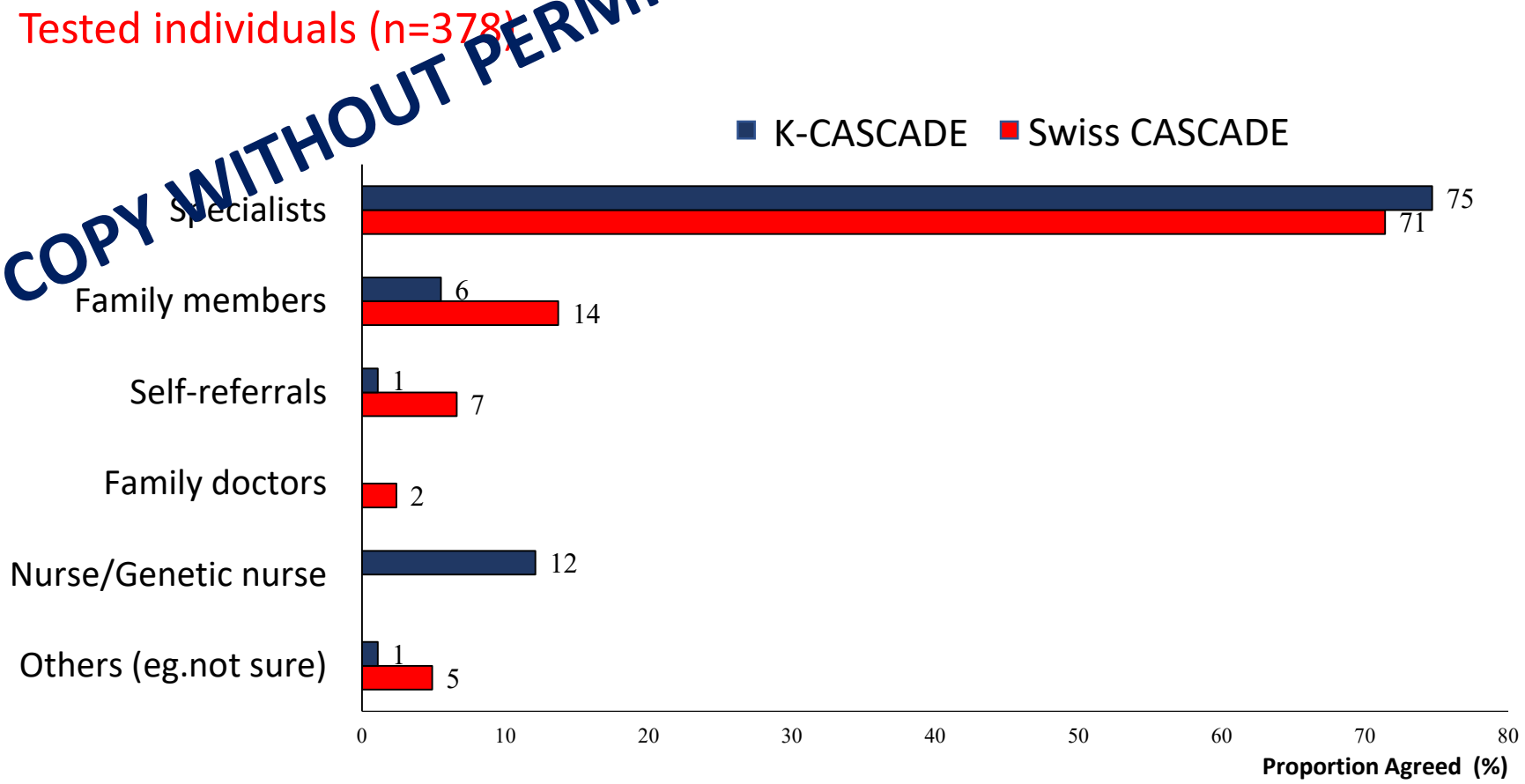
Specialists:

29% Oncologist

25% Gynecologist

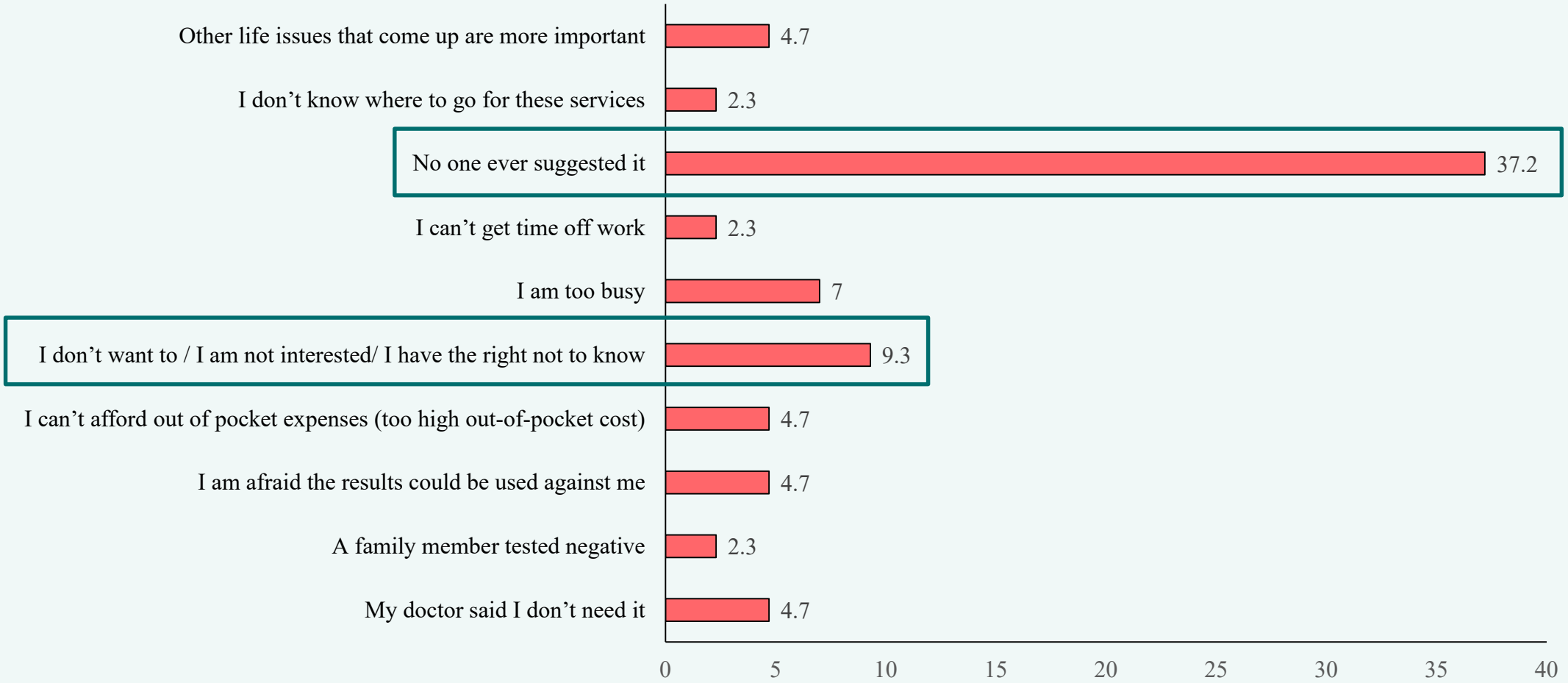
12% Medical geneticist/ genetic counselor

7% Surgeons



Swiss CASCADE Cohort

Cascade testing in untested Relatives: Reasons for not having genetic testing



SARKI ET AL. 2022 J PERS MED

Outcomes related to genomic disparities and nursing care

Outcome	Nursing care element	Examples
active **C**oping	Person- and family-centered care	Prerequisite for responsive adaptation and self-management
	Management of uncertainty	Engagement in lifelong surveillance
	Acceptance and reframing	Burden of self-disclosure and intrafamilial communication
	Supportive interventions	Receptive to hereditary disease risk
		Level of distress
intrafamilial **C**ommunication	Empower disclosure	Coaching for disclosure of test results beyond nuclear family
	Culturally-sensitive family care	Tailored approaches to reaching at-risk family members
	Skills-building interventions	Big data and biobanks

Swiss CASCADE Cohort: Nov 2021 n=287

HBOC and Lynch syndrome cases: with whom did you share genetic testing results?

- 33% with some relatives but not all
- 20% it is very difficult, don’t know how to explain test results, don’t know how to start the conversation
- 10% I feel guilty, it makes me feel sad
- 8% would rather not share results with anyone in the family

Sarki et al. 2022 Cancers; Sarki et al. 2022 J Pers Med

Swiss CASCADE Cohort: Nov 2021

Relatives potentially eligible for cascade testing vs. relatives invited

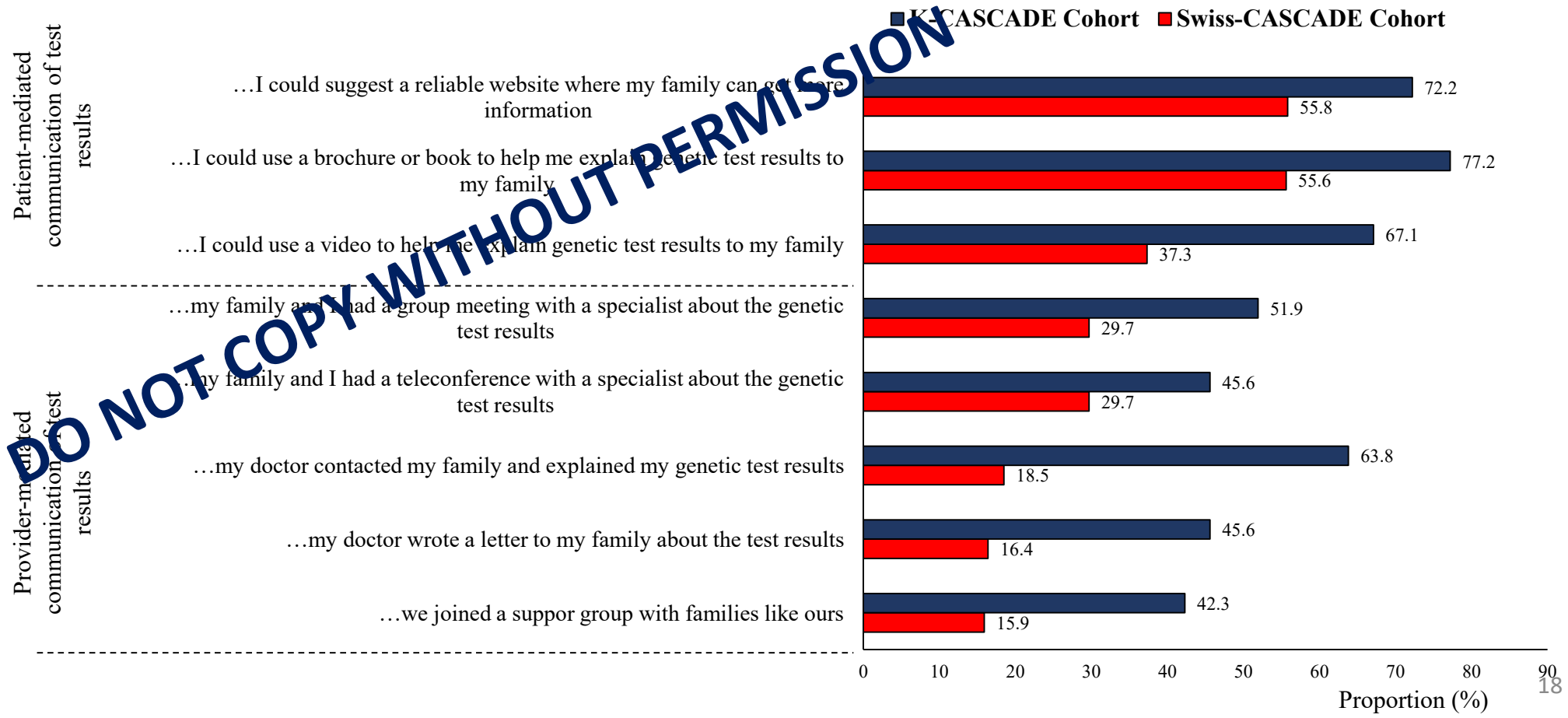
Degree of relationship	Type of relative	HBOC		LS		HBOC vs. LS [a]
		Eligible relatives	Willing to contact	Eligible relatives	Willing to contact	*p* (willing to contact)
First degree	Daughters	137	60 (43.8%)	25	9 (36.1%)	0.61
	Sons	152	75 (49.3%)	25	8 (32.1%)	0.16
	Sisters	199	69 (34.7%)	29	9 (31.3%)	0.86
	Brothers	170	58 (34.1%)	30	11 (36.7%)	0.95
	Parents	234	65 (27.8%)	47	13 (27.7%)	1
	Sum	892	327 (36.7%)	156	50 (32.1%)	0.24
Second degree	Granddaughters	32	10 (31.3%)	3	0	1 [b]
	Grandsons	23	5 (21.7%)	7	1 (14.3%)	1 [b]
	Nieces	146	41 (28.1%)	21	0	1 [b]
	Nephews	176	39 (22.2%)	20	4 (20%)	1 [b]
	Half-Sisters	23	3 (13.0)	6	2 (33.3%)	0.57 [b]
	Half-Brothers	12	1 (10%)	9	2 (22.2%)	0.58 [b]
	Aunts	229	35 (15.3%)	35	6 (17.1%)	0.97
	Uncles	205	21 (10.2%)	37	6 (16.2%)	0.40 [b]
	Grandparents	51	1 (2%)	10	1 (10%)	0.32 [b]
	Sum	897	156 (17.4%)	148	22 (14.9%)	0.52
Third degree	Female cousins	423	68 (16.1%)	47	5 (10.6%)	0.44
	Male cousins	436	47 (10.8)	76	13 (17.1%)	0.16
	Sum	859	115 (13.4%)	123	18 (14.6%)	0.81
Total		2648	598 (22.6%)	427	90 (20.8%)	0.46
Between Degree of relationship p *(Sum)* [c]		<0.01**		0.004**		
Between Gender p *(Sum)* [c]		0.17		0.60		

[a] Two proportion z- test; [b] Fisher exact test; [c] Chi-square test; ** = significant two tailed *p value* < 0.05

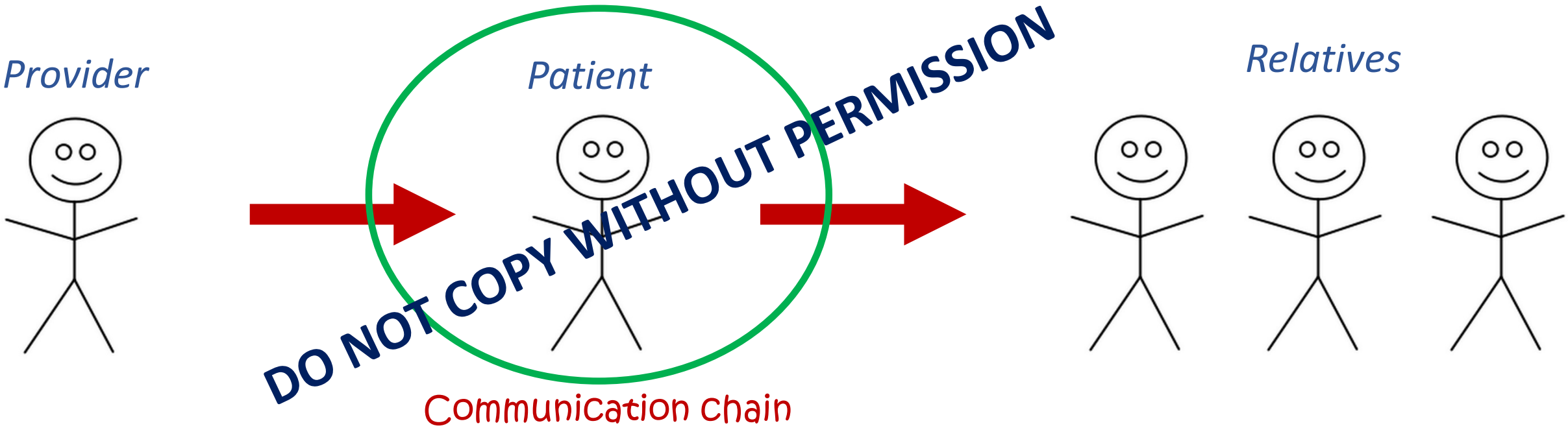
Swiss CASCADE and K-CASCADE Cohort

HBOC and Lynch syndrome cases: how can healthcare providers help you with family communication of testing results?

Tested individuals (n=378)



Understanding the patient-mediated approach



Gaps in provider-patient communication

Lack of standardization and continuity, not the best moment

Gaps in patient's genetic literacy

Lay theories

Conflicting values and loneliness in managing communication

Responsibility, self-preservation, protection of the other, respect of autonomy, harmonization of family communication

Pedrazzani et al. 2022 J Pers Med

Outcomes related to genomic disparities and nursing care

Outcome	Nursing care element	Examples
cascad**E S**creening	Identification of at-risk individuals	Target at-risk populations, cost-effectiveness of testing
	Digital technology for health communication	Tier 1 genetic conditions, public health genomics
	Referrals, resources	Expand testing to biological relatives
		Support active role of individuals with pathogenic variants
		Nurse-led interventions

Swiss CASCADE:

Sep 2017 – Jan 2022

365 index cases (50%)

276 HBOC and 89 LS

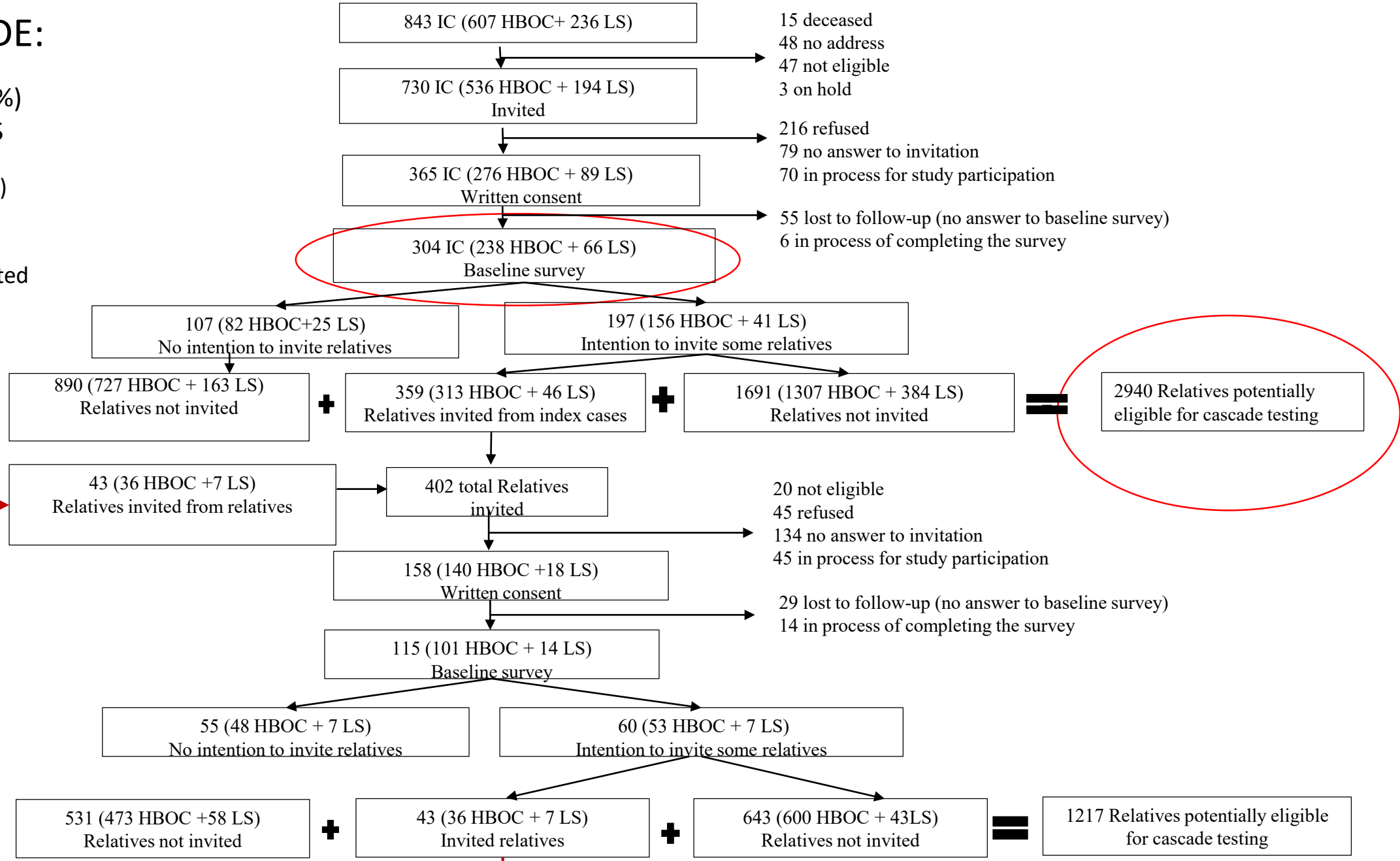
Baseline data (ongoing)

304 index cases

115 relatives (20%)

– 71 tested; 44 not tested

1st Follow-up 86.8%

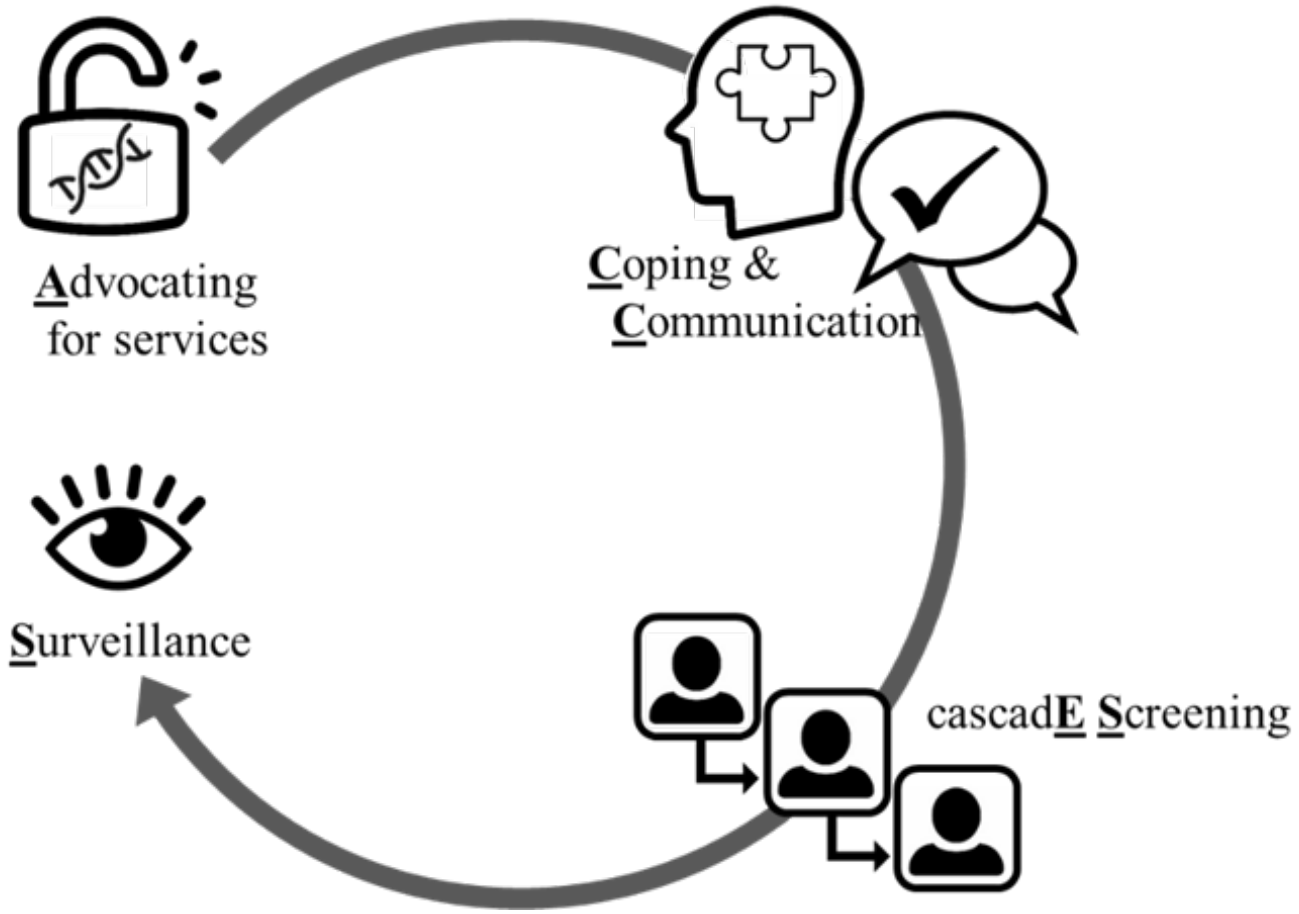


SARKI ET AL. 2022 CANCERS

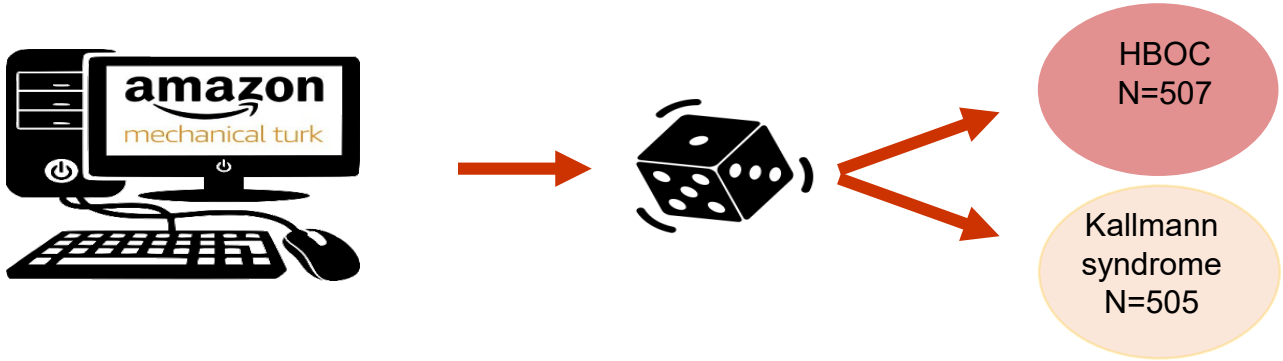
Outcomes related to genomic disparities and nursing care

Outcome	Nursing care element	Examples
lifelong **S**urveillance	Continuity of care, long-term follow-up	Continuity of care, “patient”-provider relationships
	Recurrence, reversal	Decision making for (irreversible) risk-reducing, screening,
	Lifestyle, health promotion	interpersonal relationships, reproduction, career and occupation
	Additional services	Psychosocial adaptation
		Establish high-risk clinics

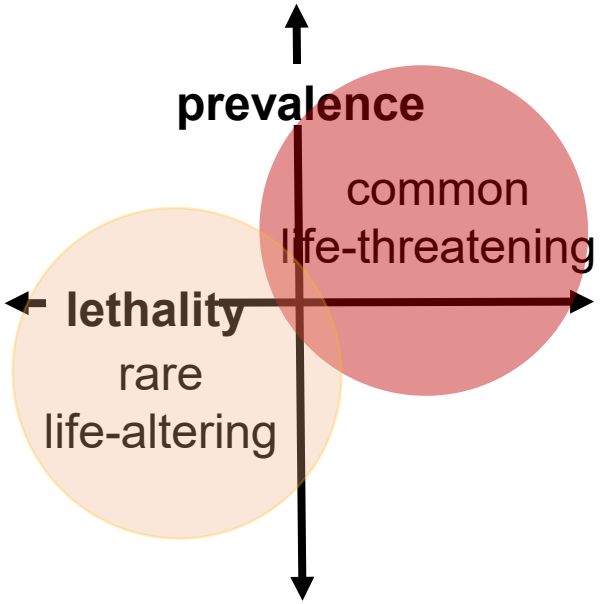
The ACCESS framework to guide nursing practice



Testing the ACCESS framework “common : rare” paradigm



Information
- clinical presentation
- diagnosis & treatment
- possible test results



No significant differences between groups
- Opting for testing
- Past experience
- Literacy/numeracy
- Satisfaction with Decision Scale
- Decision Regret Scale

Only difference in family communication and testing of relatives - expected

Dwyer et al. 2021 Genes

Conclusions

- Empower individuals, families, communities to engage and participate in technological developments
- Nurses as facilitators
- ACCESS is simple, practical guide, across the lifespan, broad applicability, generalizability

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DISCUSSION