Supplemental material

# **Supplementary Table 1. Search results from the databases**

	Search Strategy on PubMed						
S/N	Search terms	Results					
1	"Genetic Testing"[Mesh]	52,659					
	"Predisposition Test*"[Title/Abstract] OR "Predisposition Genetic Test*"[Title/Abstract] OR "Predictive Test*"[Title/Abstract] OR						
	"Predictive Genetic Test*"[Title/Abstract] OR "Predictive Screening*"[Title/Abstract] OR "Genetic Screening*"[Title/Abstract] OR						
2	"Genetic Counsel*"[Title/Abstract] OR "Genetic Service*"[Title/Abstract]	32,188					
3	#1 OR #2	77,228					
4	(("Physicians, Family"[Mesh]) OR "General Practitioners"[Mesh]) OR "Physicians, Primary Care"[Mesh]	31,077					
	"Fam* Practi*"[Title/Abstract] OR "Fam* Physician*"[Title/Abstract] OR "Family Doc*"[Title/Abstract] OR "General						
	Practi*"[Title/Abstract] OR "General Physician*"[Title/Abstract] OR "Primary Care Physician*"[Title/Abstract] OR "Primary Care						
5	Practi*"[Title/Abstract] OR "Primary Care Doc*"[Title/Abstract]	125,257					
6	#4 OR #5	137,934					
7	"Health Knowledge, Attitudes, Practice"[Mesh]	124,634					
	"Attitude*"[Title/Abstract] OR "Sentiment*"[Title/Abstract] OR "Opinion*"[Title/Abstract] OR "View*"[Title/Abstract] OR						
	"Perception*"[Title/Abstract] OR "Belief*"[Title/Abstract] OR "Feeling*"[Title/Abstract] OR "Experience*"[Title/Abstract] OR						
8	"Knowledge*"[Title/Abstract]	2,962,929					
9	#7 OR #8	2,999,417					
10	"Professional Practice"[Mesh]	265,242					
4.4	"Physician Practice Pattern*"[Title/Abstract] OR "Clinical Practice Pattern*"[Title/Abstract] OR "Physician Prescribing	4 270 054					
11	Pattern*"[Title/Abstract] OR "Practice*"[Title/Abstract] OR "Referral*"[Title/Abstract] OR "Consult*"[Title/Abstract]	1,279,051					
12	#10 OR #11	1,453,267					
13	"Education, Continuing"[Mesh]	62,498					
	"Workshop*"[Title/Abstract] OR "Educational Activit*"[Title/Abstract] OR "Training Program*"[Title/Abstract] OR						
14	"Support*"[Title/Abstract] OR "Professional Development*"[Title/Abstract]	1,937,516					
15	#13 OR #14	1,990,371					
16	#3 AND #6 AND #9	315					
17	#3 AND #6 AND #12	373					
18	#3 AND #6 AND #15	157					
19	#16 OR #17 OR #18	514					

20	#19 (Filter from 2010 - 2022)					
	Search Strategy on PsycINFO					
S/N	Search terms	Results				
S1	MA (Genetic Testing)	2,561				
	TI ((Predisposition Testing*) OR (Predisposition Genetic Testing*) OR (Predictive Testing*) OR (Predictive Genetic Testing*) OR					
	(Predictive Screening*) OR (Genetic Screening*) OR (Genetic Counsel*) OR (Genetic Service*)) OR AB ((Predisposition Testing*) OR					
	(Predisposition Genetic Testing*) OR (Predictive Testing*) OR (Predictive Genetic Testing*) OR (Predictive Screening*) OR (Genetic					
S2	Screening*) OR (Genetic Counsel*) OR (Genetic Service*))	14,954				
S3	#1 OR #2	16,623				
S4	MA (Family Physicians) OR MA (General Practitioners) OR MA (Physicians, Primary Care)	6,826				
	TI ((Fam* Practi*") OR (Fam* Physician*) OR (Family Doc*) OR (General Practi*) OR (General Physician*) OR (Primary Care Physician*) OR (Primary Care Doc*)) OR AB ((Fam* Practi*") OR (Fam* Physician*) OR (Family Doc*) OR (General					
S5	Practi*) OR (General Physician*) OR (Primary Care Physician*) OR (Primary Care Practi*) OR (Primary Care Doc*))	7,463				
S6	#4 OR #5	12,105				
S7	MA (Health Knowledge, Attitudes, Practice)	27,856				
	TI ((Attitude*) OR (Sentiment*) OR (Opinion*) OR (View*) OR (Perception*) OR (Belief*) OR (Feeling*) OR (Experience*) OR	27,030				
	(Knowledge*)) OR AB ((Attitude*) OR (Sentiment*) OR (Opinion*) OR (View*) OR (Perception*) OR (Belief*) OR (Feeling*) OR					
S8	(Experience*) OR (Knowledge*))	1,704,921				
S9	#7 OR #8	1,711,670				
S10	MA (Professional Practice)	3,502				
	TI ((Physician Practice Pattern*) OR (Clinical Practice Pattern*) OR (Physician Prescribing Pattern*) OR (Practice*) OR (Referral*) OR					
	(Consult*)) OR AB ((Physician Practice Pattern*) OR (Clinical Practice Pattern*) OR (Physician Prescribing Pattern*) OR (Practice*) OR					
S11	(Referral*) OR (Consult*))	582,019				
S12	#10 OR #11	583,422				
S13	MA (Education, Continuing)	3,084				
	TI ((Workshop*) OR (Educational Activity*) OR (Training Program*) OR (Support*) OR (Professional Development*)) OR AB					
S14	((Workshop*) OR (Educational Activity*) OR (Training Program*) OR (Support*) OR (Professional Development*))	869,843				
S15	#13 OR #14	871,475				
S16	S3 AND S6 AND S9	63				
S17	S3 AND S6 AND S12	72				
S18	S3 AND S6 AND S15	27				

S19	S16 OR S17 OR S18	100
S20	S19 (Filter from 2010 - 2022)	48
	Search Strategy on EMBASE	
S/N	Search terms	Results
1	genetic testing.mp.	48,498
	(Predisposition Testing* or Predisposition Genetic Testing* or Predictive Testing* or Predictive Genetic Testing* or Predictive Screening* or Genetic Screening* or Genetic Counsel* or Genetic Service*).ti. or (Predisposition Testing* or Predisposition Genetic Testing* or Predictive Testing* or Predictive Genetic Testing* or Predictive Genetic Screening* or Genetic Screening* or Genetic Counsel* or	
2	Genetic Service*).ab.	38,295
3	#1 OR #2	78,986
4	(family physicians or general practitioners or primary care physician).mp.	65,542
5	(Fam* Practi* or Fam* Physician* or Family Doc* or General Practi* or General Physician* or Primary Care Physician* or Primary Care Practi* or Primary Care Doc*).ti. or (Fam* Practi* or Fam* Physician* or Family Doc* or General Practi* or General Physician* or Primary Care Physician* or Primary Care Practi* or Primary Care Doc*).ab.	153,295
6	#4 OR #5	153,676
7	Health Knowledge, Attitudes, Practice.mp.	289
8	(Attitude* or Sentiment* or Opinion* or View* or Perception* or Belief* or Feeling* or Experience* or Knowledge*).ti. or (Attitude* or Sentiment* or Opinion* or View* or Perception* or Belief* or Feeling* or Experience* or Knowledge*).ab.	3,368,287
9	#7 OR #8	3,368,347
10	Professional Practice.mp.	53,808
11	(Physician Practice Pattern* or Clinical Practice Pattern* or Physician Prescribing Pattern* or Practice* or Referral* or Consult*).ti. or (Physician Practice Pattern* or Clinical Practice Pattern* or Physician Prescribing Pattern* or Practice* or Referral* or Consult*).ab.	1,613,246
12	#10 OR #11	1,646,549
13	Education, Continuing.mp.	216
14	(Workshop* or Educational Activity* or Training Program* or Support* or Professional Development*).ti. or (Workshop* or Educational Activity* or Training Program* or Support* or Professional Development*).ab.	2,220,602
15	#13 OR #14	2,220,726
16	#3 AND #6 AND #9	407
17	#3 AND #6 AND #12	528
18	#3 AND #6 AND #15	210

19	#16 OR #17 OR #18	710
20	#19 (Filter from 2010 - 2022)	490
	Search Strategy on Cochrane	
S/N	Search terms	Results
1	"Genetic Testing"[Mesh]	
	(Predisposition Testing* or Predisposition Genetic Testing* or Predictive Testing* or Predictive Genetic Testing* or Predictive	
2	Screening* or Genetic Screening* or Genetic Counsel* or Genetic Service*).ti.ab.kw	
3	#1 OR #2	
4	(("Physicians, Family"[Mesh]) OR "General Practitioners"[Mesh]) OR "Physicians, Primary Care"[Mesh]	
	(Fam* Practi* or Fam* Physician* or Family Doc* or General Practi* or General Physician* or Primary Care Physician* or Primary Care	
5	Practi* or Primary Care Doc*).ti.ab.kw	
6	#4 OR #5	
7	"Attitude"[Mesh]	
8	(Attitude* or Sentiment* or Opinion* or View* or Perception* or Belief* or Feeling* or Experience* or Knowledge*).ti.ab.kw	
9	#7 OR #8	
10	"Practice Patterns, Physicians'"[Mesh]	
	(Physician Practice Pattern* or Clinical Practice Pattern* or Physician Prescribing Pattern* or Practice* or Referral* or	
11	Consult*).ti.ab.kw	
12	#10 OR #11	
13	"Education"[Mesh]	
14	(Workshop* or Educational Activity* or Training Program* or Support* or Professional Development*).ti.ab.kw	
15	#13 OR #14	
16	#3 AND #6 AND #9 (Filter from 2010 - 2022)	19
17	#3 AND #6 AND #12 (Filter from 2010 - 2022)	34
18	#3 AND #6 AND #15 (Filter from 2010 - 2022)	22

#### Supplementary Table 2. Inclusion/Exclusion criteria

Inclusion Criteria						
Types of Genetic Testing	All types					
Paper Type	Original research, peer-reviewed journals					
	- Full text of paper available					
	- Global literature					
Time Frame	2010 –2022					
Study Population	Primary Care Physicians (PCPs), Family Physician, General Practitioner (GP)					
Focus of Paper	GPs' experiences					
	- Any or the lack thereof discussions on genetics					
	- Subjective experiences such as confidence, comfort, knowledge, barriers					
	GPs' attitudes					
	- Opinions on their role in offering clinical genetic services					
	- Awareness					
	- General views on utility of genetic testing					
	GPs' needs					
	- Educative workshops or practice policies and recommendations targeted at incorporating aspects of clinical genetic					
	services that can include taking family history, recommending and ordering tests, interpreting test results, managing					
	downstream care, and referral to clinical genetic centres					
Exclusion Criteria						
Paper Type	Commentaries, short articles, dissertations, book reviews, literature reviews, mini reviews, book chapters, editorials					
Language	Any language, other than English					
Study Population	Public, Patients, Specialist (Pediatricians, Ob-gyn, Oncologist, Geneticists) and Allied Health Professionals (nurses, health					
	educators, social workers)					
Focus of Paper	Testing of medical and clinical interventions, other than those targeted at genetic education					

Included

Studies included in review

Reports of included studies

(n = 56)

(n = 6)

Identification of studies via databases and registers Identification of studies via other methods Records removed before screening: Identification Duplicate records removed (n = Records identified from: Records identified from\*: 213) Websites (n = 0)Databases (n = 871) Records marked as ineligible Organisations (n = 0)Registers (n = 0)by automation tools (n = 0)Citation searching (n = 6)Records removed for other reasons (n = 0)Records screened Records excluded (n = 658)(n = 494)Reports sought for retrieval Reports not retrieved Reports sought for retrieval Reports not retrieved (n = 164)Screening (n = 4)(n = 6)(n = 0)Reports excluded: (n = 104)Reports assessed for eligibility Reports assessed for eligibility Reports excluded (n = 0)Paper type (n = 21)(n = 160)(n = 6)Patient/public sample (n = 18) Specialist included (n = 49) Focus on non-genetic areas (n = 14) Dupliate (n = 2)

Figure 1. PRISMA flow diagram of the study selection process

### Supplementary Table 3. Characteristics of included studies (n = 62)

Category	Studies, n (%)		
Study Type			
Quantitative	36 (58.1%)		
Qualitative	13 (21.0%)		
RCT	9 (14.5%)		
Mixed methods	4 (6.5%)		
Country			
United States	20 (32.3%)		
United Kingdom	19 (30.6%)		
Canada	11 (17.7%)		
Australia	6 (9.7%)		
Asia	4 (6.5%)		
South Africa	1 (1.6%)		
New Zealand	1 (1.6%)		
Sample Size			
< 50 respondents	21 (33.9%)		
50 – 100 respondents	12 (19.4%)		
101 – 500 respondents	21 (33.9%)		
> 500 respondents	8 (12.9%)		
Area of Focus (overlaps in articles, n>100%)			
Knowledge, experiences	50 (80.6%)		
Attitudes, views, roles	38 (61.3%)		
Needs (education, interventions)	43 (69.4%)		
Genetic Type			
General genetics	24 (38.8%)		
Oncogenetics	18 (29.0%)		
Cardiovascular	6 (9.7%)		
Pharmacogenetics (PGx)	6 (9.7%)		
Prenatal/Neonatal	4 (6.5%)		
Direct-to-consumer (DTC) testing	4 (6.5%)		
Study Quality	Avg: 4.5 (range: 4-5)		
Response rate (>50%)	11 (17.7%)		
Not reported	22 (35.5%)		

#### Supplementary Table 4. Assessment of risk of bias using the MMAT

Authors (year)	S1.	S2.	Q1	Q2	Q3	Q4	Q5
Qualitative	Are there	Do the collected data	Is the qualitative	Are the qualitative	Are the findings	Is the interpretation	Is there coherence
study	clear research	allow to address the	approach appropriate	data collection	adequately derived	of results sufficiently	between qualitative
	questions?	research questions?	to answer the	methods adequate to address the research	from the data?	substantiated by data?	data sources,
			research question?	question?		udlar	collection, analysis and interpretation?
Cusack et al.	Yes	Yes	Yes	Yes	Yes	Yes	Yes
(2021)	103	163	163	163	163	163	163
	Vos	Voc	Yes	Yes	Vos	Vas	Vac
Douma et al.	Yes	Yes	Yes	Yes	Yes	Yes	Yes
(2019)							
Fok et al.	Yes	Yes	Yes	Yes	Yes	Yes	Yes
(2021)							
Hussein et al.	Yes	Yes	Yes	Yes	Yes	Yes	Yes
(2020)							
Joshi et al.	Yes	Yes	Yes	Yes	Yes	Yes	Yes
(2020)							
Lemke et al.	Yes	Yes	Yes	Yes	Yes	Yes	Yes
(2017)							
Lemke et al.	Yes	Yes	Yes	Yes	Yes	Yes	Yes
(2020)	. 63	163			. 63	1.03	103
Mathers et al.	Yes	Yes	Yes	Yes	Yes	Yes	Yes
	163	163	163	res	163	165	163
(2010)		.,	.,,		.,		.,
McKinn et al.	Yes	Yes	Yes	Yes	Yes	Yes	Yes
(2022)							
Puzhko et al.	Yes	Yes	Yes	Yes	Yes	Yes	Yes
(2019)							
Sebastian et	Yes	Yes	Yes	Yes	Yes	Yes	Yes
al. (2022)							

Sebastian et al. (2022)	Yes	Yes	Yes	Yes	Yes	Yes	Yes
Silva et al. (2022)	Yes	Yes	Yes	Yes	Yes	Yes	Yes
Tsianakas et al. (2010)	Yes	Yes	Yes	Yes	Yes	Yes	Yes
Quantitative RCT	Are there clear research questions?	Do the collected data allow to address the research questions?	Is randomization appropriately performed?	Are the groups comparable at baseline?	Are there complete outcome data?	Are outcome assessors blinded to the intervention provided?	Did the participants adhere to the assigned intervention?
Bell et al. (2015)	Yes	Yes	Yes	Yes	Yes	Yes	Yes
Bell et al. (2014)	Yes	Yes	Yes	Yes	Yes	Yes	Yes
Carroll et al. (2011)	Yes	Yes	Yes	Yes	Yes	Yes	Yes
Houwink et al. (2015)	Yes	Yes	Yes	Yes	Yes	Yes	Yes
Houwink et al. (2014)	Yes	Yes	Yes	Yes	Yes	Yes	Yes
Houwink et al. (2014)	Yes	Yes	Yes	Yes	Yes	Yes	Yes
Wilkes et al. (2017)	Yes	Yes	Yes	Yes	Yes	Yes	Yes
Wilson et al. (2016)	Yes	Yes	Yes	Yes	Yes	Yes	Yes
Quantitative descriptive	Are there clear research questions?	Do the collected data allow to address the research questions?	Is the sampling strategy relevant to address the research question?	Is the sample representative of the target population?	Are the measurements appropriate?	Is the risk of nonresponse bias low?	Is the statistical analysis appropriate to answer the research question?

Barrow et al. (2015)	Yes	Yes	Yes	Yes	Yes	Yes	Yes
Bernhardt et al. (2012)	Yes	Yes	Yes	Yes	Yes	Yes	Yes
Bonham et al. (2010)	Yes	Yes	Yes	Yes	Yes	Yes	Yes
Carroll et al. (2019)	Yes	Yes	Yes	Yes	Yes	Yes	Yes
Carroll et al. (2016)	Yes	Yes	Yes	Yes	Yes	Yes	Yes
Challen et al. (2010)	Yes	Yes	Yes	Yes	Yes	Yes	Yes
Dunlop et al. (2010)	Yes	Yes	Yes	Yes	Yes	Yes	Yes
Evans et al. (2020)	Yes	Yes	Yes	Yes	Yes	Can't tell (No sampling frame)	Yes
Fiederling et al. (2014)	Yes	Yes	Yes	Yes	Yes	Yes	Yes
Haga et al. (2019)	Yes	Yes	Yes	Yes	Yes	Yes	Yes
Haga et al. (2012)	Yes	Yes	Yes	Yes	Yes	Yes	Yes
Haga et al. (2011)	Yes	Yes	Yes	Yes	Yes	Yes	Yes
Haga et al. (2017)	Yes	Yes	Yes	Yes	Yes	Can't tell (No sampling frame)	Yes
Kadaoui et al. (2012)	Yes	Yes	Yes	Yes	Yes	Yes	Yes

Klemenc-Ketiš	Yes	Yes	Yes	Yes	Yes	Yes	Yes
et al. (2014)							
Klemenc-Ketiš	Yes	Yes	Yes	Yes	Yes	Yes	Yes
et al. (2014)							
Laedtke et al.	Yes	Yes	Yes	Yes	Yes	Yes	Yes
(2012)							
Leitsalu et al.	Yes	Yes	Yes	Yes	Yes	Yes	Yes
(2011)							
Mainous et al.	Yes	Yes	Yes	Yes	Yes	Can't tell	Yes
(2013)						(Lack demographic	
						data)	
Marathe et al.	Yes	Yes	Yes	Yes	Yes	Yes	Yes
(2015)							
Nippert et al.	Yes	Yes	Yes	Yes	Yes	Yes	Yes
(2014)							
Nippert et al.	Yes	Yes	Yes	Yes	Yes	Yes	Yes
(2011)							
Pelletier et al.	Yes	Yes	Yes	Yes	Yes	Yes	Yes
(2020)							
Powell et al.	Yes	Yes	Yes	Yes	Yes	Yes	Yes
(2012)							
Ram et al.	Yes	Yes	Yes	Yes	Yes	Can't tell	Yes
(2012)						(Non-respondent bias	
						risk not reported)	
Rangarajan et	Yes	Yes	Yes	Yes	Yes	Yes	Yes
al. (2016)							
Richter et al.	Yes	Yes	Yes	Yes	Yes	Yes	Yes
(2013)							

Saul et al. (2017)	Yes	Yes	Yes	Yes	Yes	Can't tell (No sampling frame)	Yes
Skinner et al. (2021)	Yes	Yes	Yes	Yes	Yes	Yes	Yes
St Sauver et al. (2016)	Yes	Yes	Yes	Yes	Yes	Yes	Yes
Teng et al. (2014)	Yes	Yes	Yes	Yes	Yes	Yes	Yes
Truong et al. (2021)	Yes	Yes	Yes	Yes	Yes	Can't tell (No sampling frame)	Yes
Van Wyk et al. (2016)	Yes	Yes	Yes	Yes	Yes	Yes	Yes
Vande Perre et al. (2018)	Yes	Yes	Yes	Yes	Yes	Yes	Yes
Vansenne et al. (2011)	Yes	Yes	Yes	Yes	Yes	Can't tell (No data on non- respondents)	Yes
Yu et al. (2021)	Yes	Yes	Yes	Yes	Yes	Yes	Yes
Mixed Methods	Are there clear research questions?	Do the collected data allow to address the research questions?	Is there an adequate rationale for using a mixed methods design to address the research question?	Are the different components of the study effectively integrated to answer the research question?	Are the outputs of the integration of qualitative and quantitative components adequately interpreted?	Are divergences and inconsistencies between quantitative and qualitative results adequately addressed?	Do the different components of the study adhere to the quality criteria of each tradition of the methods involved?
Dressler et al. (2019)	Yes	Yes	Yes	Yes	Yes	Yes	Yes
Lemke et al. (2020)	Yes	Yes	Yes	Yes	Yes	Yes	Yes

Supplemental material

Mazzola et al.	Yes						
(2019)							
Schuurmans	Yes						
et al. (2019)							

## **Supplementary Table 5. Summary of key findings**

Supplemental material

S/N	Author	Title	Methods, Sample size, Country	Genetic scope	Knowledge/Experience	Attitude/Views/Roles	Needs
1	McKinn et al. 2022	Clinician views and experiences of non-invasive prenatal genetic screening tests in Australia.	Qualitative (Semi- structured interview)  4 GPs 15 F, 2 M Response rate: not reported Australia	Non- invasive prenatal screening (NIPS)	- Limited experience with high chance NIPS results - Did not often identify or voice concerns about potential harms of NIPS - Reported limited time for pre-test counselling in the context of first trimester antenatal appointments		- Some GPs suggested a mandatory training on how to discuss NIPS and disseminate the results - There also needs to be more specific guidance for GPs on the use of NIPS for screening (those currently available are focused on screening for T21), and a national system to collect routine data on NIPS requests
2	Silva et al. 2022	Introducing genetic testing with case finding for familial hypercholeste rolaemia in primary care: qualitative study of patient and health professional experience.	Qualitative (Semi- structured interview)  7 GPs 13 F, 11 M Response rate: not reported  UK	Familial hyperchole sterolaemi a (FH)	- Comfortable to refer patients with results suggesting FH or a variant of unknown significance (VUS) for specialist assessment	- Positively anticipated the value of improving identification of FH, recognising potential benefits for patients and their families' long-term health	- Sought greater understanding about interpreting and communicating the range of possible test results, and more in-depth guidance on long-term care of FH (conditions, next steps by specialists) - Anticipated a need for clearer guidance about evolving roles at the primary— secondary care interface, especially guidance on who may have what clinical responsibilities or duty of care related to genetic testing for FH, and communicating and acting on results appropriately.

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3	Cusack et	General	Qualitative (Semi-	Genomic	- Only 3 GPs attended	- Most felt their role was to	- Need for more education,
	al.	practitioners'	structured	testing	continuing professional	assist or counsel patients to	training and support
		views on	interview)		development activities on	help them understand these	resources such as clear, up-
	2021	genomics,			genetics/genomics	types of tests and results,	to-date guidelines on
		practice and	28 GPs		- Most GPs reported little	and to refer or seek advice	genomic testing; decision
		education: A	12 F, 16 M		experience with genetic or	from genetic specialists as	supports; RACGP resources;
		<u>qualitative</u>	Response rate:		genomic testing	required	patient handouts; and
		<u>interview</u>	not reported		- 3/4 reported referring	- Predict genomics to play a	opportunities to discuss
		study.			patients to genetic services	bigger part in their future	issues with a genetic
			Australia		or specialists	practice, especially for risk	specialist
					- Lack of evidence and	prediction and to inform	- Interested to learn about
					reliability of online DNA	treatment and management;	genomics with relevance to
					testing was a concern for	but a small number were	their practice
					some GPs, who stated they	uncertain.	- Prefer case studies, face-to-
					lacked confidence		face events (seminar,
					interpreting results to		workshops), online learning,
					support their patients		journals, accredited CPD
					- Challenge such longer		events, webinars and
					consultation times, cost of		podcasts.
					genomic tests		
4	Fok et al.	How practice	Qualitative (Semi-	Genetic	- Perceived level of	- Generally perceived	- GS adoption would be
		setting affects	structured	screening	confidence to conduct GS	themselves to be well-	greater if Continuing Medical
	2021	<u>family</u>	interview)		was low due to lack of	positioned to offer GS but	Education (CME) and other
		physicians'			training and knowledge	expressed ambivalence about	educational and systems
		views on	30 FPs		- Public barriers (Lack of	their current roles and	support were offered
		genetic	14 F, 16 M		control, Lower patient	competency to practise GS	
		screening: a	Response rate:		socioeconomic status and	- Some perceived that	
		<u>qualitative</u>	75%		literacy, Rigid administrative	offering GS was not core to	
		study.			infrastructure)	their scope of practice due to	
			Singapore		- Private motivations (Strong	rarity of genetic conditions.	
					longitudinal patient	- Negative patient attitudes	
					relationship, Practice	as a potential barrier	
					autonomy, Higher patient	(emotional and psychological	
					literacy)	burden)	

5	Truong et	Genetic	Quantitative	General	- 48% indicated that they	- 78% preferred either or
	al.	Referral	(Survey)	genetic	would recommend genetics	both online continuing
		Patterns and		testing	evaluation, genetic	medical education (CME)
	2021	Responses to	95 PCPs		counselling, and/or genetic	activities and online medical
		Clinical	61 F, 34 M		testing for developmental	references sites as methods
		Scenarios: A	Response rate:		delay	for obtaining genetic
		Survey of	not reported		- 71% would recommend for	information
		Primary Care			colon and uterine cancer	
		<b>Providers and</b>	US		- Concerns for financial cost	
		Clinical			to patients was the most	
		Geneticists.			common barrier	

6	Yu et al.	Preparing	Quantitative	Genomic	- 17% HK-PCPs and 40% SZ-	- 91% agreed that it was	
		genomic	(Survey)	medicine	PCPs had encountered	important to keep up to date	
	2021	revolution:			patient cases related to	with the latest information	
		Attitudes,	151 Hong Kong		genomic medicine in the past	on genetic disorders	
		<u>clinical</u>	PCPs		6 months	- 86% agreed that	
		practice, and	48 F, 103 M		- HK-PCPs were most	personalized medicine is the	
		training needs	Response rate:		confident in "obtaining	future of healthcare	
		in delivering	8%		information about genetic	- About 80% of PCPs felt that	
		genetic	258 Shenzhen		disorders from FH" and least	breast, ovarian and colorectal	
		counseling in	PCPs		confident to decide which	cancers and congenital	
		primary care	145 F, 113 M		"genetic testing should be	anomalies were conditions	
		in Hong Kong	Response rate:		done"	worth performing genetic	
		and Shenzhen,	37%		- SZ-PCPs were most	testing	
		China.			confident in referring patient	- 68% perceived ethical	
			Hong Kong,		to "a relevant specialist for	controversies associated with	
			Shenzhen, China		suspected genetic disorders"	genetic testing	
					and least confident in		
					"explaining to patients on		
					genetic testing results" and		
					"advising patients whether		
					they should do genetic test".		
					- 55% expressed insufficient		
					time during clinical		
					consultation to discuss		
					genetic issues		
					- 78% were unaware of the		
					referral pathway for patients		
					with suspected and		
					confirmed genetic disorder		

7	Joshi et al.	Primary care	Qualitative (Semi-	Genome	- Many providers felt they	- Most PCPs saw value in	- Additional resources
		provider	structured	sequencin	lacked the necessary	using GS in research for	required to facilitate GS
	2020	perspectives	interview)	g (GS)	technical expertise and skills	healthy children but diverged	testing, pretest and posttest
		on using			to convey GS results to the	in opinion on using results in	counseling, and additional
		genomic	11 FPs and 5		parents (felt unfamiliar with	primary care for children	support or training for
		sequencing in	primary care		genetic concepts and		themselves
		the care of	pediatricians		expressed discomfort with	- Proponents saw value in	- Additional resources
		<u>healthy</u>	Response rate of		interpreting and using GS	informing their patients'	incurred costs, which could
		children.	69% (11/16), 31%		results)	preventative care and	over-burden the healthcare
			(5/16)			benefiting scientific research	system
						as a whole	
			Canada			- Had more dynamic	
						definitions of actionability	
						(interventions to reduce	
						morbidity and mortality)	
						- Skeptics were driven by	
						providers' ambivalence about	
						using a research test and	
						uncertainty about what to do	
						with the result	
						- Did not feel they had a	
						professional obligation to use	
						GS results in primary care	
						- Aligned with traditional,	
						restrictive definitions of	
						actionability	

8	Pelletier et	Survey of	Quantitative	Breast and	- FPs were more likely than	
	al.	primary care	(Survey)	ovarian	gynecologists to recommend	
		physicians'		cancer	unproven ovarian cancer	
	2020	views about	134 FPs	screening	screening to a carrier but less	
		breast and	76 F, 58 M	(BRCA1/2)	likely to recommend proven	
		ovarian cancer	Response rate:		MRI screening.	
		screening for	45%			
		true BRCA1/2				
		non-carriers.	Canada			
9	Lemke et	Drimany care	Qualitative (Semi-	Genetic	- Expressed concern on the	- GWA helped increase
9		Primary care	structured		amount of time needed to	'
	al.	physician		and Wellness	discuss the alert	patient awareness of the
	2020	experiences	interview)			importance of their family
	2020	utilizing a	24 DCD=	Assessmen	recommendation due to busy	history
		family health	24 PCPs	t (GWA),	practices and patient having	- Facilitated patient-physician
		history tool	19 F, 5 M	CDS alert	difficulties answering family	discussions about disease risk
		with .	Response rate:	tool	history questions	by providing CDS alert so that
		electronic	not reported		- Lack of follow-up on the	PCPs receives specific
		health record-			testing and referrals due to	information about genetic
		integrated	US		cost, insurance concerns,	testing, personalized
		<u>clinical</u>			fear, stigma, lack of interest,	medicine services available,
		decision			and logistical issues	and next steps within the
		support: an			- Alert fatigue; CDS	health system
		implementatio			recommendations differing	- Need for more physician
		n process			from their clinical judgment;	education about the GWA
		assessment.			and technical issues	CDS recommendations

10	Evans et al.	How genomic	Quantitative	General			- Majority wanted to keep up
		information is	(Survey)	genetic			to date with genomic
	2020	accessed in		testing			medicine via online
		clinical	159 GPs				educational modules (70%);
		practice: an	Response rate:				willing to spend 30min to 1
		electronic	not reported				hour (78%) on it
		survey of UK					- More than 60% choose NICE
		general	UK				Clinical Knowledge Summary
		practitioners.					(CKS) and GP notebook for FH
							and FBC scenarios; Internet
							search engines was next most
							popular; far fewer (19%)
							access government webpage
							for information
							- Local clinical genetic
							services or seeking advice
							from specialists/secondary
							care colleagues were most
							common
11	Dressler et	<u>Implementing</u>	Mixed methods	Individualis	- None of the PCPs ordered a	- Different views on how PGz	- Individualized PGx training
	al.	<u>pharmacogen</u>	(Survey &	ed training	PGx test prior to the study as	can enhance patient care;	provided by the PM
		etic testing in	interviews)	and	they did not know/have	some prefer pre-emptive	pharmacist showed a boost
	2019	rural primary		education	access to a lab that	testing while some prefer	in physician's comfort level
		care practices:	4 PCPs	on PGx	performed PGx testing, not	testing at point of care when	with PGx testing
		<u>a pilot</u>	Response rate:	testing	seeing convincing evidence of	needed to predict response	- Shift in barriers from lack of
		<u>feasibility</u>	not reported		clinical utility, and not feeling	to drug	expertise, lack of comfort to
		study.			confident to interpret and		more practical issues of costs,
			US		apply results in treatment		and issues related to
					decision		electrical medical records
					- Pricing continues to be		- Clinical interpretative
					concern and barrier for		summaries provided by the
					physicians		PM Pharmacist were very
							helpful

12	Schuurma	Feasibility of	Mixed methods	Expanded	- Over time they developed a	- Most GPs were positive	- All GPs interviewed said
12	ns et al.	couple-based	(Survey &	carrier	routine for conducting the	about combining ECS pre-test	they felt able to provide the
	iis et ai.	expanded	interviews)	screening	counselling, which reduced	counselling with GPC	pre-test counselling mainly
	2019	carrier	interviews)	(ECS)	the time required for	- GPs considered themselves	because of the training,
	2019	screening	10 GPs	(LC3)	preparation and counselling	as the most suitable	supervision and additionally
		offered by	(interview), 116		itself		provided materials such as
			•			providers for a population-	I -
		general	GPS (checklist)		- GPs did not experience any	based ECS couple-test.	study checklist as a practical
		<u>practitioners.</u>	Response rate:		barriers in communicating	- ECS-provision as standard	guidance
			90%		the normal results or to	care by all GPs might not be	
			<del>-</del>		referring any couples at	feasible because not all may	
			The Netherlands		normal risk to Clinical	be able to keep up with	
					Genetics for additional pre-	technological advances; only	
					or post-test counselling	motivated GPs willing to do	
						so should be trained to	
						provide ECS	
						- Some were resistant to	
						additional workload due to	
						busy practices	
13	Douma et	<u>Information</u>	Qualitative	Lynch	- Generally followed the	- Felt responsible for	- Like to have rapid access to
	al.	<u>exchange</u>	(Interview)	syndrome	patient's request to be	referring patients for follow-	information and information
		<u>between</u>		(LS)	referred for genetic	up care and also for providing	specifically tailored for GPs.
	2019	patients with	6 GPs		counselling and rely on the	support.	- GPs appreciated the letter
		<u>Lynch</u>	Response rate:		cancer family history that	- Did not perceive this to be	from the genetic HP;
		syndrome and	not reported		patients provide on their own	their responsibility	generally, they only had
		their genetic			initiative		contact with the
		and non-	The Netherlands		- Provide very little		gastroenterologists via
		genetic health			explanation about LS to their		letters.
		<u>professionals:</u>			patients at the time of		
		<u>whose</u>			referral, as they lacked the		
		responsibility?			knowledge		
					- Several GPs were not		
					regularly informed by GEs		
					about the endoscopic		
					surveillance, while others		
1	ĺ				reported to receive letters or		
1					were unsure about whether		

					they were informed by the gastroenterologists.		
14	Vande	Role of the	Quantitative	BRCA1/2	- 81% collected the family	- Many (72.4%) felt not	- 27.5% were trained during
	Perre et al.	general	(Survey)		history	included or that they had a	their initial training to care
	2018	practitioner in the care of	58 GPs		- 24% considered they had sufficient knowledge of the	minor role (31%) in the care of their patients	for patients with a BRCA1/2 mutation
	2010	BRCA1 and	24 F, 34 M		indication criteria for	- 72.4% saw their role in	- Only 11.8% of the GPs
		BRCA2	Response rate:		genetics consultation	caring for these patients is	attributed their knowledge
		mutation	38.2%		- 69.7% considered that they	limited to psychological	on the subject to the referral
		<u>carriers:</u>			were not able to answer	support and to motivate	guidelines of the French
		<u>General</u>	UK		patients' questions about	relatives to undergo	national cancer institute
		practitioner			BRCA1/2 guidelines	screening (70.7%).	(INCa).
		and patient			- 75.9% were not familiar		- 32.8% reported receiving a
		perspectives.			with the criteria for referring		letter from the geneticist
					patients to cancer genetics		- 79.6% are interested in
					consultations		training

15	Wilkes et	Increasing	RCT (survey)	BRCA,		- Interactive web-based CME
	al.	<u>confidence</u>		genetic		was more effective at
		and changing	121 PCPs - 60	testing,		improving knowledge and
	2017	behaviors in	intervention; 61	perinatal		shared decision making
		primary care	control			behaviors but had a small
		providers	40.5% F, 59.5% M			effect on attitudes and
		engaged in	Response rate:			minimal impact on clinical
		genetic	3.5%			behaviours on ELSI
		counselling.				discussions
			US			- Intervention showed
						greater increase in
						knowledge, more satisfied
						with educational materials
						and more confident in their
						ELSI genetic knowledge and
						skills
						- Self-efficacy improved in
						both groups; intervention
						showed significantly higher
						improvements

16	Lemke et	Primary care	Qualitative (Semi-	Pharmacog	- PGx testing results were	- PGx testing could help	- Undergoing direct access
	al.	physician	structured	enetic	used to adjust patient	individualize medication	PGx testing themselves was a
		experiences	interview)	(PGx)	medications to increase	treatments for their patients	useful teaching tool and that
	2017	<u>with</u>		testing	effectiveness and reduce side	- Utility of PGx testing was	it was helpful for them to
		integrated	15 PCPs		effects	helpful for patients to	have first-hand knowledge of
		pharmacogen	60% F, 40% M		- Lack of understanding of	potentially avoiding	the testing and resulting
		omic testing in	Response rate:		the pharmacogenomics test	medication side effects and	process
		a community	not reported		report and how to interpret,	guide decision-making for	- Desire for clarification on
		health system.			not adequately prepared to	patients starting a new	the results report and
			US		communicate complex	medication	preferred certain formats for
					results	- Using PGx direct access	results display as well as a
					- Delay to receive results was	testing can foster increased	paper copy of the results
					a barrier in providing timely	patient autonomy and	- More PGx education (such
					patient feedback	satisfaction (more efficient	as in-services, case studies,
					- Time constraints as a	and save the additional	and online training) to guide
					challenge and the need for an	costs), and assurance on	on how to address cost and
					in-office follow-up	medication plan	insurance issues with
					appointment to discuss	- Few did not think PGx	patients
					results	testing was useful in their	- Further training specific to
						patient population now but	results report interpretation
						will be more valuable in the	- Interested in receiving both
						future	provider and patient
						- High cost and lack of	education materials
						reimbursement for patients	(colourful pamphlets, etc.)

17	Haga et al.	Primary care	Quantitative	Pharmacog	- 58% reported ordering	- 83% believed that	- Awareness on PGx was
		providers' use	(Survey)	enetic	genetic testing for disease	pharmacists would have	gained from professional
	2017	of pharmacist		(PGx)	diagnosis one-time or two-	some or a large role in	meetings, drug or laboratory
		support for	12 PCPs	testing	times per year	delivering PGx; 75% believed	representative, publications,
		delivery of			- All 12 GPs indicated that	that geneticists/genetic	CME learning, grand rounds
		pharmacogen	US		they did not feel well	counselors would have some	or point-of-care notification
		etic testing.			informed about genetic	or a large role in delivering	- 75% prefer to learn about
					testing in general nor about	PGx testing	PGx through grand rounds or
					PGx testing specifically	- 42% believed that the	other in-house seminars
					- 2 GPs felt comfortable to	physician who ordered a PGx	- 92% indicated having some
					discuss PGx testing prior to	test should communicate test	assistance in interpretation
					ordering test, 3 GPs felt	results to the patient	would increase likelihood to
					somewhat comfortable to	- 5 GPs believed that either	order a PGx test
					discuss PGx test results, 3	the ordering physician, a	- 63% consulted pharmacist;
					GPs felt comfortable using	genetic counselor or a	providers who did not
					PGx test to inform treatment	pharmacist could	consult the pharmacist did so
					decisions	communicate PGx results	because they did not feel
							they needed pharmacist's
							input or they did not have
							time
							- All agreed that having a
							pharmacist available is
							helpful (meet patients, more
							learning opportunities with
							pharmacist, mail written
							summary of test results)
18	Wilson et	Supporting	RCT (survey)	Hereditary	- FPs' intentions were lower	- Taking family history seen	
	al.	genetics in		breast and	for 'making a risk	positively as a normal activity	
		primary care:	96 PCPs	ovarian	assessment' (perceived as	for FPs; but a proportion	
	2016	investigating	Response rate:	cancer	the most difficult, saw no	were sceptical if this should	
		how theory	76.8%	(HBOC)	value, felt no pressure or	be part of their practice due	
		<u>can inform</u>			confidence to do it) than for	to lack of confidence to take	
		<u>professional</u>	Canada		the other two behaviours	FH	
		education.			('taking family history' and		
					'making referral')		

19	St Sauver	Integrating	Quantitative	Pharmacog			- 52% did not expect or know
	et al.	<u>Pharmacogen</u>	(Survey)	enomics			how to use pharmacogenetic
		omics into		(PGx)			information in future
	2016	<u>Clinical</u>	90 PCPs				practices
		<u>Practice:</u>	Response rate:				- Of those that received alert,
		<u>Promise vs</u>	57%				53% felt that alerts were
		Reality.					confusing, irritating or
			US				difficult to find additional
							information
							- Only 30% changed their
							prescription in response to
							PGx alert
20	Van Wyk	<u>Knowledge</u>	Quantitative	Hereditary	- GPs have limited	- Most GPs are interested to	- Important resources
	et al.	<u>regarding</u>	(Survey)	cancer	knowledge about basic	learn more or become more	includes: CME (86%),
		basic concepts		(HBOC, LS,	concepts of hereditary	involved in referring at-risks	discussion with colleagues
	2016	of hereditary	61 PCPs	FAP)	cancers and local genetic	patients appropriately.	(82%), guidelines (82%),
		cancers, and	Response rate:		facilities available.	- Most agreed that patients	published data, journals
		the available	31.1%		- Majority were unsure how	should have counselling	(88.5%)
		<u>genetic</u>			to perform risk assessments;	before testing.	
		counselling	South Africa		only 36% would refer to		
		and testing			appropriate cancer genetic		
		services: A			services		
		survey of			- 65% felt that they were not		
		general			sufficiently qualified and		
		<u>practitioners</u>			equipped to provide genetic		
		<u>in</u>			counselling; and agreed that		
		Johannesburg,			genetic counsellors (100%),		
		South Africa.			medical geneticist (85%) and		
					oncologist (68%) were more		
					qualified		

Supplemental material

21	Marathe et	<u>General</u>	Quantitative	Genetic	- 51.4% feel confident in	- 100% agreed that it is	- Education was needed
	al.	Practitioners'	(Survey)	cardiac	educating patients with GCDs	important to educate	through monthly newsletter
		knowledge		diseases	but 29.3% were unsure	patients about their genetic	or in the form of creating
	2015	and use of	144 GPs	(GCDs)	- 39.6% were not confident to	condition	clinical pathways to assist in
		genetic	73 F, 71 M		answer patient's questions	- 95.1% also agree that it is	referring appropriately
		counselling in	Response rate:		about GCD	important to educate family	
		managing	21%		- 56% did not feel confident	members about genetic	
		patients with			with the knowledge they	conditions	
		genetic	Australia		have regarding GCDs but 56%	- 94.3% mentioned	
		<u>cardiac</u>			were confident with their	cardiologist or specialist as	
		<u>disease in</u>			knowledge in appropriately	being most important in the	
		non-			managing GCDs in their	team of GCD care providers	
		<u>specialised</u>			clinical practice	for guidance, 2 GPs also saw	
		settings.			- 76.1% routinely educated	it as the cardiologists' role to	
					patients and their relatives	refer	
					- 86.7% had heard about the		
					Tasmanian Genetic		
					Counselling Service but		
					52.8% knew little about the		
					service provided		
					- Variations in referral: 37.3%		
					said that they sometimes		
					referred, 26.8% did routinely		
					refer, 14.8% did not routinely		
					refer, and 10.6 % only		
					referred if the patient asked		
					for it		

22	Barrow et	Improving the	Quantitative	Lynch	- 77.8% had no previous	- 49.2% did not feel this was	- 74.6% highlighted the lack
	al.	uptake of	(Survey)	syndrome	experience of referring a	part of their role	of supporting literature to
		<u>predictive</u>		(LS)	patient/family with	- 90.5% felt that patients	facilitate the discussion
	2015	testing and	63 GPs		suspected LS to the Regional	themselves had the most	
		<u>colorectal</u>	Response rate:		Genetics Service, 79.4% were	responsibility for adhering to	
		screening in	29.2%		unclear which patients	the recommended screening	
		<u>Lynch</u>			should be referred for	guidelines although 50.8%	
		syndrome: a	UK		investigation	identified this as part of their	
		<u>regional</u>			- 73.0% were unaware of the	role also	
		primary care			Regional Lynch Syndrome	- Shared responsibility among	
		survey.			Registry	healthcare professionals,	
					- 61.9% had no experience of	including the Regional	
					discussing cancer risk, 38.1%	Genetics Service, the	
					had no experience discussing	gastroenterologist/colorectal	
					screening recommendations	surgeon and GP, with most	
					- 87.3% did not feel confident	responsibility for screening	
					to discuss the details of LS	lying with the screening	
					- 57.1% had concerns over	centres.	
					confidentiality which would		
					prevent them from		
					approaching		
					at-risk relatives		
					- Barriers includes lack of		
					knowledge and time		
					constraints (41.3%)		

23	Bell et al.	Impact of a	RCT (survey)	BRCA		- Intervention had minimal
		randomized				impact on practices to offer
	2015	controlled	121 PCPs - 60			genetic counselling but with a
		educational	intervention; 61			few exceptions:
		trial to	control			- Intervention-physicians
		<u>improve</u>	40.5% F, 59.5% M			were more likely to explore
		<u>physician</u>	Response rate:			genetic counseling benefits;
		<u>practice</u>	3.5%			advise for a test decision
		<u>behaviors</u>				after counselling; and inform
		<u>around</u>	US			that postiive results would
		screening for				indicate increased risk of
		inherited				prostate cancer for male
		breast cancer.				relatives
						- Intervention-physicians
						were less like to ask about
						Ashkenazi heritage
						- Specific questions about
						cancers in the family,
						including ovarian, breast, and
						prostate cancers, were not
						usually asked.
						- Cost, implications of
						treatment, and limitations of
						current genetic testing were
						not usually discussed.
						- Majority offered referral to
						geneticists

24	Teng et al.	Attitudes and	Quantitative	Cancer	- 87.5% have referred	- 84 % wanted more
		knowledge of	(Survey)	genetic	patients for cancer genetic	information
	2014	medical		testing	testing (GPs referred 1 in 790	
		practitioners	32 GPs		patients)	
		to hereditary	Response rate:		- 60% correctly estimated the	
		cancer clinics	25%		cost of the first family	
		and cancer			member (proband) to	
		genetic	Australia		undergo cancer genetic	
		testing.			testing	
					- 20% correctly estimated	
					turnaround time for	
					routine cancer genetic	
					testing, and 30% for urgent	
					cancer genetic testing	
					- Wide discrepancy between	
					the self-reported	
					GP referral rate (87.5 %) and	
					the actual referral rate	
					calculated from patient files	
					(12.5 %)	
					- Reasons for not referring:	
					no treatment, no patient	
					request for it	

25	Houwink	<u>Effectiveness</u>	RCT (survey)	Oncogenet		- Case-based oncogenetics
	et al.	<u>of</u>		ics		education can achieve
		oncogenetics	56 GPs (38			sustained improvement (3
	2014	training on	intervention, 18			mths after the training)
		<u>general</u>	control group)			- Positive results for active
		<u>practitioners'</u>	41 F, 15 M			and interactive sessions,
		<u>consultation</u>	Response rate:			single-group and smaller-
		skills: a	64% (56/88)			group sessions
		<u>randomized</u>				- Participating GPs seemed to
		controlled	The Netherlands			be more comfortable
		<u>trial.</u>				incorporating oncogenetics
						into patient consultation
						skills (high applicability skills)
26	Houwink	<u>Sustained</u>	RCT (survey)	Oncogenet		- Online genetics CPD module
	et al.	effects of		ics		can result in sustained
		<u>online</u>	44 GPs			improvement of genetics
	2014	<u>genetics</u>	39 F, 5 M			knowledge
		education: a	Response rate:			- More than 90% applied
		<u>randomized</u>	55%			newly acquired knowledge at
		controlled trial				least once a month
		<u>on</u>	The Netherlands			- Self-reported applicability
		oncogenetics.				aspects focused indicates
						that the G-eCPD mainly
						improved genetics
						knowledge rather than skills

а	al.	communicatio	(C)				
1		Communicatio	(Survey)		cancer family history is raised	Germany, the Netherlands	
		n, predictive			in a consultation "at least	and the UK considered	
2	2014	testing and	1197 GPs		once a week"/"once a	practice responsibility should	
		management	Gender reported		month"	be "to provide support after	
		in France,			- GPs from Germany (76.6%)	breast cancer testing"	
		Germany, the	UK (France,		and France (74.3%) reported	- GPs from France ascribed to	
		<u>Netherlands</u>	Germany, the		that they would always take a	the following tasks: "explain	
		and the UK:	Netherlands and		family history whereas only	the inheritance pattern of	
		general	the UK)		36.0% of the Dutch and	familial breast cancer",	
		<u>practitioners'</u>			40.1% of the British GPs	"inform about breast cancer	
		and breast			reported always taking FH.	genetic risk for the relatives",	
		surgeons'			- Majority reported that they	"inform about breast cancer	
		<u>current</u>			"always"/"frequently"	genetic testing", "provide	
		practice and			provide risk assessment	support after breast cancer	
		<u>preferred</u>				genetic testing", and "inform	
		<u>practice</u>				about possible management	
		<u>responsibilitie</u>				options available after the	
		<u>S.</u>				results of breast cancer	
						genetic testing".	
						- GPs from all countries	
						unanimously agreed that	
						"disclose breast cancer	
						genetic test results to the	
						patient" should be	
						undertaken by a genetic	
						specialist.	
I I	Fiederling	Consideration	Quantitative	Family	- 53% reported that they		- 57% did not feel there is a
e	et al.	of family	(Survey)	history of	only ask for FHC in general,		need for standardized tool to
		history of	25.05	cancer	but not for a specific cancer		collect information on FHC
2	2014	cancer in	35 GPs	(FHC)	site. Those who noted asking		- 60% feel that there is a
		<u>medical</u>	Response rate:		for specific cancer sites most		need for further information
		routine: a	70%		frequently asked for a family		or guidelines regarding
		survey in the			history of breast or CRC		preventive counselling of
		<u>primary care</u>	Germany		- 97% would screen according		individuals with a FHC
					to general guidelines and		- Most prefer either

29 Mainous AG 3rd et al. Description of perception of integration integration into practice: a CERA study.  30 Laedtke et al. Description of al. Description of into practice: a CERA study.  30 Laedtke et al. Description of into practice: and information into practice: and into practic	17%) or tool (14%) for unselling
AG 3rd et al. physicians' perception of genetic testing and and into practice: a CERA study.  30 Laedtke et al. physicians' awareness 2012 and knowledge of the Genetic linformation Non-  AG 3rd et al. physicians' perception of genetic testing perception of genetic testing and and and the physicians' perception of integration into practice: a CERA study.  30 Laedtke et al. physicians' awareness 2012 and knowledge of the Genetic linformation Non-  AG 3rd et al. physicians' perception of genetic testing to the strain their knowledge on available genetic testing even though they anticipate on future clinical practice. Of the have substantial impact of GT to have substantial impact on future clinical practice.  AS 1. Canada, US  DTC  AS 2012 and physicians' (Survey) awareness of GINA the Genetic linformation on mination their knowledge of the Genetic linformation Non-  Non-  AS 383 FPS (GINA)  AS 2008 (GINA)  AS 2008 (GINA)  AS 2008 confident in their knowledge on available genetic testing even though they anticipate to test patient's risk for disease but less so to determine suitable treatment for patient - Self-perceived knowledge was positively associated with prediction on impact of GT, and importance of GT curriculum - 58.1% felt that DTC was more likely to harm patients' general health decisions  AS 2012 and physicians' (Survey) and prior awareness of GINA and claimed a basic understanding - Most common concern for	
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30 Laedtke et al. physicians' (Survey) awareness and awareness and but had limited knowledge, knowledge of the Genetic Information Non-    Response rate: Information Non-   Response rate: Act of Information Non-   Response rate: Act of Survey (GINA)   CINA	
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the Genetic Information Non- Response rate: Act of 2008 understanding - Most common concern for	
Information   26.9%   2008   understanding   Non-	
Non- (GINA) - Most common concern for	
Discrimination US discrimination was on life	
Act (GINA). insurance (49.6%)	

31	Haga et al.	Primary care	Quantitative	Pharmacog	- 51.4% strongly or	- 64.5% agreed that PGx	- Preferred methods to
		physicians'	(Survey)	enetic	somewhat disagreed that	testing is or will soon be a	educate PCPs were CME (in-
	2012	knowledge of		(PGx)	they felt well-informed about	valuable tool to predict risk	person courses) 36.5%,
		<u>and</u>	40.58% Fam	testing	genetic testing	of adverse events or	training in residency 15.5.
		<u>experience</u>	medicine, 58.21%		- 73.0% did not feel that their	likelihood of effectiveness	- Most PCPs learned about
		<u>with</u>	internal med,		genetics training adequately	- Most (62.9%) believed that	PGx through journals (46.9%)
		pharmacogen	1.21% other		prepared them to	they should have primary	or professional meetings,
		etic testing.	34.04% F, 65.96%		appropriately order or use	responsibility for making	CME, or grand rounds
			M		genetic tests.	patients aware of a PGx test	(46.61%).
			Response rate:		- 43.7% strongly or somewhat	- 57.5% believed it was their	
			15% (597)		disagreed that they felt	responsibility as a primary	
					comfortable ordering a test	care provider to discuss PGx	
			US		to predict disease	test results with the patient	
					susceptibility		
					- Only 13% felt well-informed		
					about the role of PGx testing		
					in therapeutic decision-		
					making		

32	Powell et	Primary care	Quantitative	DTC	- 61.3% had never heard or	- Among the 63 respondents	
	al.	physicians'	(Survey)	testing	read about DTC genetic	(42.6%) who thought that	
		awareness,			testing	testing was clinically useful	
	2012	<u>experience</u>	382 PCPs		- Among those that had read,	when formulating medical	
		and opinions	115 F, 263 M		common sources of	management plans, most	
		of direct-to-	Response rate:		information were medical or	frequently endorsed benefits	
		consumer	16.2%		scientific journals (35.1%),	were the ability to: 1) offer	
		genetic			television (33.1%), a	screening tests at an earlier	
		testing.	US		newspaper article (28.4%)	age to individuals at an	
					and the Internet (27.0%)	increased risk (82.5%, n = 52),	
					- Older PCPs (41 and above)	and 2) offer screening tests	
					were almost twice as likely to	more frequently to	
					be aware of DTC genetic	individuals who are found to	
					testing than younger PCPs.	be at an increased risk	
					- 81.1% had never discussed	(81.0%, n = 51).	
					DTC tests with a patient or	- Among the 85 respondents	
					had a patient bring in results	who thought that it is not	
					of DTC genetic tests	clinically, reasons endorsed	
					- 33.8% felt DTC genetic test	were 1) no guidelines exist to	
					results were likely to	reduce or alleviate the risk	1
					influence the care of patients	for many diseases (80.0%,	
					in their practice	n = 68), 2), it is too difficult to	1
					- 85% did not feel prepared	interpret what the results	
					to answer their patient's	mean regarding patient care	
					questions regarding DTC	(58.8%, n = 50), 3), it will	
					genetic testing	cause more patient anxiety	
						(51.8%, n = 44), 4), they	
						would not change a patient's	
						management based on DTC	
						testing (35.3%, n = 30)	

33	Ram et al.	<u>General</u>	Quantitative	DTC	- Only half of respondents	- Respondents were	
		practitioner	(Survey)	testing	had heard about DTC genetic	ambivalent on benefits of	
	2012	attitudes to			testing.	DTC but agreed with risks and	
		direct-to-	113 GPs		- GPs who had received	barriers presented; those	
		consumer	49 F, 64 M		training disagree that DTC is a	without training emphasised	
		genetic testing	Response rate:		useful service of healthcare	on proposed benefits while	
		in New	38%		- Lack of knowledge,	those with training	
		Zealand.			experience and time were all	emphasised on proposed	
			New Zealand		considered barriers to GPs	risks.	
					providing genetic counselling	- Genetic specialist was	
						highlighted as the most	
						appropriate to provide	
						counselling.	
34	Kadaoui et	Breast cancer	Quantitative	Breast	- For women aged 35 to 49		
	al.	screening	(Survey)	cancer	years, more than 80% of		
		practices for			physicians reported using		
	2012	women aged	460 GPs		practices deemed adequate,		
		35 to 49 and	247 F, 206 M		except for instruction in BSE		
		70 and older.	Response rate:		and referral for genetic		
			36%		counseling (60% and 54%).		
					- For women 70 years of age		
			Canada		and older with GLE, only 50%		
					of general practitioners		
					prescribed screening		
					mammography.		
					- For the 70 years and older		
					age group without GLE, for		
					whom screening is not		
					indicated, nearly half of		
					physicians continued to		
					perform CBE and more than		
					one-third continued to		
					review family history		

35	Haga et al.	Genomic risk	Quantitative	Genetic	- 45% strongly or somewhat	- 53% expressed concerns	- Preferred educational
		profiling:	(Survey)	testing	strongly agreed that they felt	about life and long-	resources to learn about
	2011	attitudes and			well-informed about genetic	term/disability insurance	genomic risk profiling: CME
		use in	79% Internal		testing	discrimination, 50% about	courses (69%), medical
		personal and	med, 19.1%		- 52% strongly or somewhat	health insurance	journals (57%), professional
		clinical care of	family medicine,		strongly agreed that they	discrimination, 43% about	medical meetings (53%), and
		primary care	1.9 other		would feel comfortable	confidentiality, 41% about	educational programs offered
		<u>physicians</u>	14.6% F, 85.4% M		ordering genetic testing for	inadequate knowledge of	by testing companies (47%)
		who offer risk	Response rate:		disease susceptibility	testing, and 36% indicated	
		profiling.	44%		- Significant association	they did not believe testing	
			(167)		between feeling well-	would provide useful	
					informed and feeling	information	
			US		"comfortable" ordering a		
					genetic test - those who felt		
					well-informed were more		
					likely to feel comfortable		
					(78.6%) than those who did		
					not feel well-informed		
					(29.8%).		
					- 49% did not believe that		
					their genetics training was		
					adequate.		

36	Carroll et	GenetiKit: a	RCT (survey)	Hereditary		<ul> <li>Multifacted educational</li> </ul>
	al.	randomized		breast and		intervention could
		controlled trial	80 PCPs - 47	ovarian		significantly improve referral
	2011	to enhance	intervention; 33	cancer		decisions, to be more
		delivery of	control	(HBOC)		consistent with guidelines
		genetics	49 F, 31 M			and, instil greater confidence
		services by	Response rate:			in core genetics
		<u>family</u>	64%			competencies
		physicians.				- Intervention-physicians
			Canada			showed lower decisional
						difficulty and higher
						appropriate referral decisions
						score; higher confidence
						across all competencies
						- Among intervention-
						physicians: materials (Gene
						messenger) were generally
						useful; 93% would like to
						continue receiving
						information, 93% would
						recommend to their
						colleagues; 76% said that
						practice changed 'a little'
						with 9% stating changing 'a
	.,	5 ' 1'	0		5 65	lot'
37	Vansenne	Providing	Quantitative	Neonatal	- Few GPs were aware of	
	et al.	genetic risk	(Survey)	screening	primary goals of reporting	
	2011	information to	131 GPs	(Sickle	carriers was identify and	
	2011	parents of newborns	131 GPS 59 F, 72 M	Cell)	guide reproductive decisions of parents.	
		with sickle cell	Response rate:		- Barriers includes intrinsic	
		trait: role of	49% unadjusted		(lack of clinical experience)	
		the general	4970 unaujusteu		and extrinsic (rarity of sickle	
		practitioner in	The Netherlands		cell)	
		neonatal	The Netherlands		- Majority reported the lack	
		screening.			of specific clinical experience	
		Joi Cerning.		l	or specific cliffical experience	

					and knowledge on disease and inheritance	
38	Nippert et al. 2011	Confidence of primary care physicians in their ability to carry out basic medical genetic tasks-A European survey in five countries-Part 1.	Quantitative (Survey)  1168 GPs 1454 F, 2226 M Response rate: not reported  UK (France, Germany, the Netherlands, and the UK)	Genetic tasks	- 64.4% were not confident to perform basic genetic tasks (take and identify FH, identify and explain autosomal family patterns, estimate risk, recognise malformations, provide psychosocial counselling, identify patient support groups, identify relevant information, identify specialist genetic services) - 19.3% did not receive any genetic training and 61.1% had only undergraduate training 34.2% have at least one patient per month with a genetic condition and 17.9% report more than one patient contact due to a genetic condition per week	- 12.8% attended CME/CPD courses in genetics

39	Bonham et	<u>Patient</u>	Quantitative	Genetic	- Majority of physicians		
	al.	physical	(Survey)	screening	reported that they would not		
		characteristics			offer genetic screening but		
	2010	and primary	968 PCPs - 495		race was a significant factor		
		care physician	saw black patient,		in their decision		
		decision	473 saw white		- Physicians were 1.5 times		
		making in	patient		more likely to offer genetic		
		preconception	324 F, 668 M		screening to black patient in		
		genetic	Response rate:		clinical vignette compared to		
		screening.	10%; unadjusted		white patient.		
					- 88% reported age as a		
			US		factor that influenced their		
					decision to offer screening		
40	Tsianakas	Offering	Qualitative (in-	Antenatal	- Organisational barriers:	- GPs saw the benefits of	- Materials and trainings
	et al.	antenatal	depth interview)	sickle cell	lack of time, best left to	offering antenatal screening	were found to be helpful for
		sickle cell and		and	midwives to inform patients,	in primary care, as early	future screenings
	2010	<u>thalassaemia</u>	25 PCPs - 17	thalassae	inability to understand	screening will provide	
		screening to	intervention; 8	mia (SC&T)	English	additional options for	
		pregnant	control	screening		pregnant women therefore	
		women in	Response rate:			improving healthcare.	
		primary care:	not reported				
		a qualitative					
		study of GPs'	UK				
		experiences.					

41	Sebastian	Widening the	Qualitative (Semi-	Secondary		- PCPs in both groups	- Without actionability, PCPs
	et al.	lens of	structured	genomic		approached SFs through the	described that patients were
		actionability:	interview)	findings		lens of actionability: by	only left with the potential
	2022	A qualitative	,	(SFs)		looking for clinical actions	harms of learning SFs
		study of	15 FPs (3 patient,	(0.0)		that could be taken based on	(anxiety from not knowing
		primary care	12 hypothetical			this information	what to do, potential for
		providers'	patient)			- Did not consider all SFs to	unnecessary follow-up
		views and	10 F, 5 M			be beneficial because they	investigations with physical
		experiences of	,			did not perceive all SFs to be	and psychological patient
		managing	Canada			actionable.	harm, escalating cost)
		secondary				- All PCPs saw the benefit of	, , ,
		genomic				medically actionable and	
		findings.				pharmacogenomic SFs such	
						as referrals, alternative	
						medications or dosages, and	
						entering this information	
						prominently into the EMR for	
						future clinical decision	
						making	
42	Sebastian	<u>Challenges</u>	Qualitative (Semi-	Secondary	Challenges related to clinical	- Most providers described	- Innovative practice
	et al.	and practical	structured	genomic	practice:	feeling responsible for	solutions - clinical decision
		solutions for	interview)	findings	- Lack of time to manage SFs	incorporating secondary	support tools, web-based
	2022	managing		(SFs)	in a busy practice (time	findings into their practice,	patient portals, chatbots
		<u>secondary</u>	15 FPs (3 patient,		required to discuss results)	but a limited capacity to	- Comprehensive letter and
		genomic	12 hypothetical		- Lack of	manage these finding	report - make results easier
		findings in	patient)		familarity/knowledge with		to understand and navigate
		primary care.	10 F, 5 M		genomics terminology and		- New EMR feature to store
					genomic tests (knowledge		genomic information
			Canada		challenges)		
					- Technology (EMR) - inability		
					to appropriately store		
					genomic information		

43	Skinner et	<u>Interpretation</u>	Quantitative	Genetic	- FPs are more likely to		
	al. 2021	and	(Survey)	testing	misinterpret or mismanage		
		management			basic genetic information		
		of genetic test	67 FPs		- 49% of FPs were unable to		
		results by	Response rate:		correctly estimate carrier		
		Canadian	not available		status for an autosomal		
		<u>family</u>			recessive condition, although		
		physicians: a	Canada		they tended to err on the		
		<u>multiple</u>			side of overestimating risk in		
		choice survey			this scenario		
		<u>of</u>			- 69% of the responses to the		
		performance.			scenario were inappropriate		
					with microarray testing		
					replacing karyotype		
44	Hussein et	<u>Is family</u>	RCT (focus group,	Family	- FH not collected	- Mismatched in attitudes	
	al.	history still	in-depth	history	consistently and	and practice where taking FH	
		underutilised?	interview)		systematically but only if GPs	is an important part of	
	2020	Exploring the			felt it was necessary or	clinical assessment to identify	
		views and	25 PCPs		relevant to patients either:	hereditary conditons; GPs	
		experiences of	18 F, 7 M		- Proactively for health	have a role to play but	
		primary care	Response rate:		screening; prevalent	approaches varies	
		doctors in	not reported		multifactorial conditions		
		Malaysia.			(diabetes, cardiovascular		
			Malaysia		disease); newly registered		
					patients		
					- Reactively when specific		
					genetic symptoms appears		
					- GPs seldom draw pedigree		
					as EMR is not user-friendly;		
					difficult and time consuming;		
					patients having difficulty		
					recalling their FH		

45	Lemke et	Primary care	Mixed methods	Genetic	- Most PCPs (74.3%)	- PCPs highlighted the value	- Suggested the need for
	al.	physician	(Survey &	testing	reported feeling concerned	of genetic testing in	both patient and provider
		experiences	interviews)		about the privacy of their	identifying risk to detect and	educational resources such as
	2020	<u>with</u>			patients' genetic test results	prevent disease in patients	patient education handouts
		integrated	17 PCPs		and the potential for health	and their families	(78.6%) and physician
		population-	(interview)		(60.3%) and life (91.5%)	- 77% somewhat or strongly	reference sheets (78.5%)
		scale genetic	70 PCPs (survey,		insurance discrimination	agreed that the genetic	- 56.5% were satisfied overall
		testing: A	67.3%)		- 52.8% feel confident	testing program is useful to	with the DNA-10K program
		mixed-	35 F, 34 M		explaining the risks and	change their current	- Additional education on
		<u>methods</u>			benefits of genetic testing to	management of patients'	medical management options
		assessment.	US		their patients - cancer risk	care	for patients with a positive
					(42.9%), cardiac risk (27.2%)	- 81.4% agreed that the	result (88.4%) and clinical
					and PGx (32.8%)	genetic testing program has	testing guidelines (86.6%).
					- Confidence to explain	value in identifying the need	
					results was slightly higher	for increased disease	
					than their reported ability to	screening and supporting	
					articulate clear next steps	patient care management	
					- 86.8% reported that the	(69.6%)	
					genetic testing program has		
					increased their workload		
					- Only 28.9% agreed that they		
					have received adequate		
					training to offer genetic		
					testing in their practice		
					- 40.0% reported being		
					confident in their knowledge		
					of genetics, their ability to		
					explain genetic concepts		
					(47.1%) and results to		
					patients (34.8%) and their		
					ability to respond to patient		
					questions about genetic		
					technologies (27.9%)		

46	Carroll et	Informing	Quantitative	Genetic	- Lack knowledge and	- FPs see a role for	- Resources: Very few could
	al.	Integration of	(Survey)	testing	confidence in GM skills	themselves in taking FH,	find useful information
		Genomic			needed	identifying genetic condition,	regarding genetic services
	2019	Medicine Into	361 FPs		- Involvement in key tasks to	making appropriate referrals,	with regards to their own
		Primary Care:	Response rate:		deliver traditional GM:	supporting patients	practice.
		<u>An</u>	26.4% adjusted		Majority were highly involved	- Mixed attitudes (somewhat	- Useful resources includes
		Assessment of			in some aspects of traditional	optimistic and cautious about	local genetic clinic contact
		<u>Current</u>	Canada		GM tasks (identifying;	current clinical benefits).	information, genetic referral,
		Practice,			referrals; providing support)	- Mixed attitudes: Majority	testing and guidelines; most
		Attitudes, and			but less so in others	expect advances in GM to	popular suggestion for
		<u>Desired</u>			(evaluating results; discussion	improve patient's health	integration was contact
		Resources.			on benefits, risk and	outcomes but fewer than half	(telephone/fax/email) or
					limiations)	agreed it was important to	buddy system with
					- Low confidence: Self-	learn about personalised	geneticists
					reported confidence on GM	patient care based on	
					skills were moderate to low -	genomics; it was their	
					participants who indicated	responsibility; genomics as an	
					interests were more likely to	exciting part of practice	
					have a higher confidence		
					score; agree in advances of		
					GM; seeing it as their		
					responsibility		

47	Haga et al.	Primary care	Quantitative	DTC	- 62% did not receive any	- Positive experience with a	- Preferred mode of
	2019	physicians'	(Survey)	testing	type of formal education in	novel application or service	education for genomic
		knowledge,			genomic medicine	may improve future	medicine is online CME
		attitudes, and	82 FPs, 48		- 42% had referred 1-3	knowledge acquisition	programs (42%), followed by
		<u>experience</u>	Internal Medicine		patients for a genetic	regarding this specific test	professional meetings (21%),
		with personal	64 F, 66 M		consultation in the past year	and related applications, as	and in-person CME such as
		genetic	Response rate:		- 44% have never ordered a	well as potentially alter	grand rounds (18%).
		testing.	not reported		genetic test	practice behaviors	
					- Top 3 concerns were the	- Attitudes improved	
			US		lack of established clinical	significantly following testing	
					practice guidelines (72%),	regarding confidence in	
					uncertain clinical utility	discussing results of DTC	
					(65%), and personal lack of	genetic testing, knowledge	
					knowledge to interpret the	about discussing risks,	
					information (56%)	benefits and results of DTC	
					- 92% had none or minimal	genetic testing as well as	
					knowledge of GWAS	patients' ability to	
					- 61% had minimal	understand their results and	
					knowledge about when and	perceived benefit	
					how to integrate genomic		
					medicine into practice		
					- 59% reported that testing		
					experience improved their		
					knowledge of genomic		
					medicine a little		

48	Puzhko et	<u>Health</u>	Qualitative	Breast	- Time restriction due to the	- PCPs agreed that	- Use public campaigns,
	al.	professionals'	(Interview)	cancer	lack of time at a typical	implementation of this new	invitation perceived as being
		perspectives			appointment was among the	program could be beneficial	issued by the government
	2019	on breast	~11 PCPs		most important concerns	for women.	would add to the chances of
		cancer risk			- Major concern of PCPs was		being accepted
		stratification:	Canada		the interpretation of the		- More evidence that the risk
		<u>Understandin</u>			meaning of the new breast		stratification model is
		g evaluation			cancer risk stratification		beneficial and provide
		of risk versus			approach and its advantages		justification of the value
		screening for					- Suggested engaging a nurse
		disease.					other trained personnel, or
							the creation of a helpful
							online tool
							- Being able to use a
							validated tool for guiding
							screening practices, rather
							than being influenced by
							women's anxiety, would be
							beneficial

49	Saul et al.	Survey of	Quantitative	Genetic	- 99% collected information	- 84.8% agreed that PCPs	- 3/4 were interested in CME
		family history	(Survey)	testing	about the family health	have a duty to warn families	programs having to do with
	2017	taking and			history	about risks in the family.	genetics in primary care
		genetic testing	349 PCPs		- 88.3% felt confident in their	- 71.8% felt there are	
		in pediatric	224 F, 124 M		ability to determine	situations in which it is the	
		practice.			the need for further	role of the PCP to provide	
			US		evaluation based on the	genetic testing and	
					results of the FH	evaluation	
					- 50.6% refer many or most		
					of their patients identified as		
					at-risk for a genetic related		
					disorder to geneticists or		
					other specialist		
					- 95% had referred patients		
					for genetic consultation		
					- Lack of training on genetic		
					risks and choosing		
					appropriate tests (53.1%),		
					inadequate time during		
					typical office visit to interpret		
					tests (48.9%), lack of training		
					in genetic interpretation		
					(60.2%), and lack of		
					guidelines for care		
					management (57.4%).		

50	Rangarajan	Knowledge	Quantitative	Familial	- Significant shortfall in	- 82% saw GPs as most	- 69.2% prefer interpretative
	et al.	<u>and</u>	(Survey)	hyperchole	awareness, knowledge and	effective in the early	comments and alerts from
		awareness of		sterolaemi	practices on FH among GPs;	detection of FH	labs to highlight at-risk
	2016	<u>familial</u>	133 PCPs	a (FH)	role of primary care in FH has		patients
		<u>hypercholeste</u>	Response rate:		not been adequately defined		
		<u>rolaemia</u>	77.37%		- Overall knowledge on FH		
		among			among GPs was low (40.6%		
		registered	India		aware of international		
		medical			guidelines; 12.8% aware of		
		<u>practitioners</u>			preventive, management and		
		in tamil nadu:			referral services of FH)		
		Are they			- 41.4% were unsure if they		
		suboptimal?.			had FH patients; FH is		
					undiagnosed in the		
					community		
51	Carroll et	The Gene	Quantitative	Genetic			- 92% indicated that their
	al.	Messenger	(Survey)	testing			practice would be changed or
		<u>Impact</u>					improved by at least one of
	2016	Project: An	1402 FPs				the rated Gene Messengers
		<u>Innovative</u>	842 F, 560 M				- 79% of the Gene Messenger
		Genetics	Response rate:				ratings indicated FPs had
		Continuing	7.4%				learned something new
		Education					- 88% were satisfied with
		Strategy for	Canada				Gene Messengers, 76% found
		Primary Care					this method of pushed emails
		<u>Providers.</u>					useful for learning about
							genetics and found Gene
							Messengers useful for clinical
							practice
							- 94% wanted to continue to
							receive them
							- FPs commented that this
							method was an ideal way to
							stay up to date in an evolving
							field such as genomics, and
							that the email push "forced"

							them to learn about genomics topics that they might not have sought out
52	Klemenc-	<u>Family</u>	Quantitative	Hereditary	- Only 50% feel competent to	- More than 70% believe	
	Ketis et al.	physicians' management	(Survey)	cardiomyo pathy	interpret genetic risks; 25% will give genetic testing	taking FH is part of their job but 70% also believe that	
	2014	of genetic	271 FPs	(HCM)	information; 6% will interpret	ordering and discussing	
		aspects of a	75.6% F, 24.4% M	, ,	results	genetic test/implications is	
		<u>cardiac</u>	Response rate:		- Younger FPs more willing to	not part of their job	
		disease: A	27.1%		include genetic tasks in	- FPs believe it is a family	
		scenario-	LIIZ		everyday practice	responsibility to inform their	
		based study from Slovenia.	UK		- FPs with more genetic education more willing to	relatives of risk but almost 70% would choose not to	
		itom siovema.			refer patients to	respect patients' wishes and	
					genetic/cardiovascular	inform relatives themselves	
					assessment		

53	Bell et al.	Detecting	RCT (Case-	Familial		- A telephone call from a
		familial	historical control	hyperchole		chemical pathologist to the
	2014	hypercholeste	study)	sterolaemi		requesting GP of a patient at
		rolaemia in		a (FH)		high risk of FH significantly
		<u>the</u>	82 GPs			improves FH detection and
		community:	(intervention), 83			specialist referral rates in
		Impact of a	GPs (control)			addition to interpretative
		telephone call				comments
		<u>from a</u>	Australia			
		<u>chemical</u>				
		pathologist to				
		the requesting				
		general				
		<u>practitioner.</u>				
54	Richter et	<u>Variants of</u>	Quantitative	BRCA 1/2	<ul> <li>24% 'always/consistently'</li> </ul>	
	al.	<u>unknown</u>	(Survey)		mention VUS as a possible	
		significance in			test result upon referral	
	2013	BRCA testing	21 FPs			
		impact on risk	Response rate:			
		perception,	44%			
		worry,	_			
		prevention	US			
		<u>and</u>				
		counseling.				

55	Bernhardt	Incorporating	Quantitative	DTC	- Only 50% of respondents	- 40% agreed that such	
	et al.	direct-to-	(Survey)	testing	ordered a genetic test more	results would be helpful in	
		consumer			than once a year, and only	patient management	
	2012	genomic	315 Internal		16%	- 49% of respondents agreed	
		<u>information</u>	medicine, 187		ordered tests once a week or	that this kind of testing will	
		into patient	Fam Med.		more.	be commonplace in the next	
		care: Attitudes	98 F, 401 M		- 58% of respondents	5 years (respondents who	
		<u>and</u>	Response rate:		reported feeling confident in	ordered genetic tests at least	
		experiences of	23.3%		interpreting genetic test	once a month were	
		primary care			results	significantly more likely to	
		physicians.	US		- 20% had no genetics	agree)	
					education, while 56% had a	- 43% of respondents	
					genetics course in medical	indicated they would be likely	
					school	or very likely to change the	
					- 22% felt their training in	management of the	
					genetics was sufficient to	hypothetical patient	
					work with their patients who	(approximately one-third did	
					have had genetic testing	not mention the disorders	
						they would address and gave	
		1				nonspecific response)	
56	Dunlop et	'Start the	Quantitative	Family			- 30% reported that they had
	al.	conversation':	(Survey)	history			heard about the campaign
	2212	the New	400.05				through one or more sources:
	2010	South Wales	138 GPs				the newsletter of the Division
		(Australia)	57 F, 53 M				of General Practice (60%),
		family health	Response rate:				mail or e-mail (48%), an
		history	23%				article in 'Australian Dr'
		<u>campaign.</u>	Acceturalia				(40%), general media
			Australia				including television interview
							and newspaper articles (40%), and other which
							included patient, family, or
							friends (5%).
							- Only 18% reported that they
							had seen or currently had
							one or more of the campaign
						<u>l</u>	one of filore of the campaign

							resources: pads of 'Tips on collecting a family health history' (n = 20), the FHH collection tool 'My Family Health Record' (n = 22), and the poster (n = 14)
57	Challen et al.	General practitioner	Quantitative (Survey)	Hereditary cardiac	- 38% willing to explain inheritance; 28% willing to	- Although 61% consider it part of their role to take a FH,	
	di.	management	(Survey)	disease	carry out other tasks	far fewer (less than 25%)	
	2010	of genetic	1,168 PCPs	4.50450	- German, Swedish and UK	would be willing to discuss	
		aspects of a	404 F, 764 M		more likely to do initial tasks	specific genetic tests or their	
		cardiac	Response rate:		(taking FH) while French	implications. This results also	
		disease: a scenario-	28.6%		would either carry out most tasks or refer for the entire	vary according to the specific country context.	
		based study to	France 236,		genetic package		
		<u>anticipate</u>	Germany 251,				
		providers'	Netherlands 254,				
		practices.	Sweden 262, UK 165				
			103				
			UK				

58	Houwink	Effect of	RCT (survey)	Oncogenet			- For G-eCPF, self-reported
	et al.	comprehensiv		ics			genetic consultation skills
		<u>e</u>	92 GPs - 42 in G-				and consideration of referral
	2015	oncogenetics	eCPF; 50 in live				to clinical genetics centres
		training	training program				increased after one year but
		interventions	Response rate:				number of regional referrals
		for general	52% (G-eCPD);				did not change
		practitioners,	57% (live training)				- 88% of GPs who attended
		evaluated at					live training session more
		<u>multiple</u>	The Netherlands				frequently considered
		<u>performance</u>					referring patients to genetic
		<u>levels.</u>					centres than those who
							attended online CPD (64%)
59	Klemenc-	<u>Family</u>	Quantitative	Genetic	- Majority of FPs received	- FPs expressed clear role in	
	Ketis et al.	physicians'	(Survey)	testing	education from	genetics and perceived	
		self-perceived			undergraduate studies	genetics to be highly	
	2014	importance of	271 FPs		- O6674% reported having	important	
		providing	205 F, 66 M		contact with patients with	- More than 90% felt that it	
		genetic test	Response rate:		genetic disease weekly	was their duty to discuss	
		information to	27.1%			genetic testing issues with	
		patients: a	UK			their patients; especially	
		cross-	UK			positive and negative test	
		sectional study from				results, and risk of inheritance	
		Slovenia.				- FPs expressed lower	
		Sioverna.				interests on ethical issues	
60	Leitsalu et	Giving and	Quantitative	Genetic	- PCPs do not show great	- There was postive attitudes	- Majority agree that training
	al.	withholding of	(Survey)	screening	confidence in their own	among PCPs regarding the	program on GT is necessary
	ai.	information	(Survey)	Jercennig	ability to discuss genetic test	introduction of genetic	program on a 13 necessary
	2012	following	64 PCPs		results with patients and	information into clinical	
		genomic	Response rate:		families but tend to provide	practice and receiving	
		screening:	41.54%		risk information for specifc	additional training in	
		challenges			conditions regardless of	genomics, but varies based	
		identified in a	UK		circumstances	on patient.	
		study of			- Majority feel comfortable to	- Majority believe that	
		primary care			talk about basic genetics and		

		physicians in Estonia.			take FH but most were not comfortable to talk about inheritance patterns - False security, unnecessary anxiety were two common concerns	predictive genetic testing will improve healthcare	
61	Mathers et al. 2010	Family history in primary care: understanding GPs' resistance to clinical genetics qualitative study.	Qualitative (Indepth Interview)  21 GPs 12 F, 9 M Response rate: not reported  UK	Genetic testing	- GPs also admit that they are not confident about their genetic knowledge - Routine use of FH for clinical decision making is distinguised from genetic conceptualisation; FH is an integral part of general practice and not just for diagnosis or risk-assessment but also psychosocial dimensions - GPs expressed concern over being right, being updated with evidence, and making appropriate management decisions	- Although genetic concepts are part of GP practice, they are made distinct from genetics and genetic practice; not identified as core component of their practice Genetics/genetic practice not perceived to have significant impact on their practice; which are seen as rare, complex and specialist	- Call for education, training and guidelines; but need is not echoed by all

62	Mazzola et	Primary care	Mixed methods	Exome	- Knowledge scores were	- Even though PCPs may not	
	al.	physicians'	(Survey &	sequencin	positively associated with	fully understand ES, majority	
		understanding	interviews)	g (ES)	comfort score to perform	found ES beneficial for their	
	2019	and utilization			genetics tasks and referrals;	patient's care and identified	
		of pediatric	27 PCPs		more recent genetic training	and recognise positive clinical	
		<u>exome</u>	Response rate:		showed higher knowledge	utility of ES results	
		sequencing	12.6%		and confidence scores	- PCPs look to GHPs to	
		results.				communicate results and	
			US			manage follow up directly	
						with patients; 74% of PCPs	
						agree that its family	
						responsibility to follow up on	
						results	