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Genetic research on hereditary breast cancer in Latin America and the Caribbean: a systematic review

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ABSTRACT

To identify the countries with studies on hereditary breast cancer (CaMaH) in Latin America and the Caribbean (LAC) and the frequency of genes with pathogenic variants (VP).

Method: We used the PRISMA 2020 framework as a reference to conduct a systematic review of the literature on the topic, using an explicit method to collect and synthesize the findings of individual studies, including original scientific articles on CaMaH, published between January 2002 and July 2021.

Results: We found 112 genetic studies on CaMa in 21 of 48 countries and territories of LAC (44%), 17 had not been included in previous reviews and eight were first publications,

mainly from Central America and the Caribbean. More than half of the countries have not published studies on CaMaH and a third have no studies on the subject. The most frequently reported genes with VP were: BRCA2 (39.1% in Puerto Rico), BRCA1 (25% in the Bahamas), Tp53 (8.6% in Brazil), and PALB2 (4.3% in Barbados).

Discussion: The study of CaMaH in LAC remains uneven, but there are important advances in several countries and studies. The BRCA1/2 genes are the most studied, followed by PALB2 and TP53. There is heterogeneity in the design, type of population and genes studied, limiting the establishment of general conclusions. The frequency of genes with VP is diverse in each country and region. It is important to expand genetic screening in the at-risk population and strengthen national care programs.

INTRODUCTION

Hereditary or familial breast cancer (CaMaH) accounts for about 10% of all CaMa, with germline mutations occurring in susceptibility genes, e.g., BRCA1, BRCA2, TP53, CHEK2, PTEN, ATM, and PPM1D. These genes can be of high and low penetrance and interact with multiple genes and environmental factors (Mahdavi et al., 2019).

The countries of Latin America and the Caribbean (LAC) are characterized by a genetically diverse population of native and immigrant populations, with the proportion of each genetic component unknown. It is necessary to know the characteristic mutations in each population to generate genetic profiles that contribute to improving the diagnosis and treatment of CaMaH. There are few studies on CaMaH reporting for two decades foundational pathogenic variants (VP) associated with increased risk.

Asthon-Prolla and Regla (2014) conducted the first systematic review on three foundational genes (BRCA1, BRCA2, and TP53) identifying 22 studies in six countries: Brazil (11), Chile (3), Colombia (3), Costa Rica (2), Mexico (2) and Venezuela (1), covering the period 2002-2013. This study was followed by that of Ossa and Torres (2016), who reviewed the founder and recurrent mutations in BRCA1 and BRCA2, reporting 20 studies, between 2007 and 2015, in ten countries: Brazil (5), Mexico (3), Colombia (3), Chile (2), Argentina (2), Costa Rica (1), Cuba (1), Peru (1), Uruguay (1) and Venezuela (1).

Jara et al. (2017) also studied mutations in BRCA1/2 and other susceptibility genes in the Central and South American population, reviewing 47 studies in twelve countries (which), in 5956 individuals found 190 different VPs for BRCA1/2. In that same year, Chavarri et al (2017) studied the genetic evaluation of the risk of CaMa in twelve LAC countries and Hispanic women in the United States, finding 23 studies with information on BRCA1/2, in the period 2007-2016: Mexico (5), Colombia (3), Argentina (2), Bahamas (2), Brazil (2), Chile (2), Costa Rica (2), Cuba

(1), Peru (1), Puerto Rico (1), Uruguay (1), Venezuela (1) and Hispanics in the United States (1). They found that BRCA1 was the most frequent VP gene, except for Costa Rica, Cuba, Puerto Rico, and Uruguay where BRCA2 was more common.

The most recent by Urbina Jara et al. (2019) reviewed 81 studies from eleven LAC countries between 2000 and 2019 finding that most focused on BRCA1/2 genes: Brazil (32), Chile (14), Mexico (12), Colombia (6) Argentina (4), Peru (3), Puerto Rico (3), Uruguay (3), Costa Rica (2), Cuba (1) and Venezuela (1).

While previous studies provided an initial overview of the CaMaH situation in certain countries in the LAC region, there were still areas to be explored, therefore, the objective of the study is to identify the countries that have studied CaMaH throughout the LAC region and the frequency of genes reported with VP.

METHOD

We used the PRISMA 2020 framework as a reference to conduct a systematic review of the literature on the topic, using an explicit method to collect and synthesize findings from individual studies that address the formulated question: Which LAC countries have studied CaMaH? and What are the most frequent genes reported with VP? The events studied are the existence of scientific publications and the most frequent genes with VP reported in cases of CaMaH, and the outcome is the proportion of countries reporting the frequency of such genes.

The inclusion criteria were a) gene frequency studies with CaMaH VP; (b) published between January 2002 and July 2021; c) original scientific articles (descriptive and analytical) and systematic reviews, both paper and electronic; and d) conducted in one of the 48 LAC countries or territories. The exclusion criteria were a) genetic studies not associated with CaMaH; (b) case studies and non-scientific publications; and c) published outside the period studied.

As a source of information, we searched the PubMed and SciELO databases (EMBASE, LILACS, and BIREME). Specifies the date each resource was last queried. Regarding the search strategy, terms were used such as “hereditary breast cancer”, “familial breast cancer”, Latin America, Caribbean, (a specific search was made with the name of each of the 48 countries or territories of LAC), “genetic studies”, “BRCA1, BRCA2”, “PALB2”, “TP53”, “non-BRCA genes”, “pathogenic variants”. Titles and abstracts were reviewed.

In the selection process, the two researchers performed the review and selection independently at the beginning and then jointly. In case of discrepancy, the inclusion/exclusion criteria were revised together and the article was included if relevant. The bibliography obtained from each article was reviewed to identify other articles. Bibliographic records, tables dis aggregated by country, and the frequency of genes identified with VP were made.

Regarding the data extraction process: one reviewer (JG) collected the data from each publication, which was validated by the second reviewer (MC), reviewing the most relevant aspects of the original publication. The studies were grouped by country, and within each group by frequently reported genes. In terms of outcomes, they were (a) the existence or absence of molecular studies on CaMaH by country; and, b) the frequency of genes reported with VP. We calculated proportions for studies in each country and extracted relevant data for descriptive tables on the most studied studies.

To synthesize the data, LAC countries were classified into three groups: with genetic studies on CaMa (Group 1), with non-genetic studies on CaMa (Group 2), and countries without studies on CaMa (Group 3). Only studies on CaMaH were included in the synthesis. A flow chart of the review of the information, a table of general characteristics of the publications by country, and a frequency table of genes reported with VP were elaborated. To reduce the risk of bias, the authors, working independently, identified, reviewed, and selected studies, using diverse sources.

Regarding the assessment of bias in publication, we acknowledge that studies have variable methodological qualities and that there are risks inherent in the inclusion of each primary study. Being mainly descriptive studies there are risks of bias derived from this type of study. The aspects that were controlled are initially defined in the inclusion criteria, which were original studies, focused on the research question, all the tables included in the reports were reviewed and, when possible, the additional tables provided in the links. The inclusion, review, and selection of the studies were carried out by the two researchers, independently, until a consensus was reached.

RESULTS

We found 8,161 studies on CaMa in general, 2,213 molecular studies on CaMa, and 112 genetic studies on CaMaH, which were selected for inclusion in the review and summarized in the flowchart (see Figure 1). According to the titles and abstracts, some molecular studies met the inclusion criteria, but upon review, they were found not to be specific to CaMaH.

The included studies are specific for CaMaH and provide information on the frequency of genes found with VP. The studies were descriptive, family, case-control, and cohort studies. The type of patients included patients with CaMa, CaMaH, and variable age groups. The differences by specific patient groups were not analyzed, only frequency ranges of the genes with VP by country were presented, which is a limitation for the interpretation and cause of heterogeneity in the results.

There may be bias in study identification, although we explored various databases and sources, as well as reference screening of selected studies. We conducted comprehensive searches by country and type of genes analyzed to increase the certainty of the results.

Geographical disparity of CaMaH research in LAC

While studies on CaMa were found in 31 of the 48 LAC countries (65%), only 21 (44%) have published genetic studies on CaMa (Group 1), 10 (21%) have conducted non-genetic CaMa research (Group 2) and 17 (35%) found no study on CaMa (Group 3). (Table 1)

Group 1: One or more studies on CaMaH

Brazil: With 36 articles, it is currently the country at the forefront of CaMaH research. As of 2004, thirteen studies on BRCA1/2 mutations have been reported (Lourenco et al, 2004; Dufloth et al. 2005a; Duflothy et al. 2005b; Gomes et al. 2007; da Costa et al. 2008; Esteves et al. 2009; Ewald et al 2011; Dillenburg et al 2012; Ewald et al 2016; Fernandes et al 2016; Maestro et al 2016; de Oliveira et al 2016; Alemar et al 2017; Palmero and cabbage 2018. As of 2007, twelve articles on TP53 are identified with Achatz et al, 2007; followed by Assumpcao et al 2008; Palmero et al. 2008; Garritano et al. 2010; Giacomazzi et al. 2011; Rodriguez et al. 2011; Gomes et al 2012; Cury et al 2014; Giacomazzi et al 2013, Giacomazzi et al 2014; Almeida et al 2016; Andrade et al 2016; Hahn et al 2018. Abud et al, in 2012, reported the first study on CHECK2. Comprehensive studies on BRCA1/2, CHECK, and TP53 were conducted by Carraro et al. 2013; Felix et al. 2014; Palmero et al. 2016; and Cipriano et al. 2019. Possuelo et al, 2013, studied polymorphisms of GSTM and GSTT1. In the search for rare variants in patients negative for common variants, the studies of Torrezan et al. 2018 stand out; Silva et al. 2014; de Souza et al. 2018, in which the mutation in ATR was identified.

In Chile, we found 20 studies since 2002, ten of them related to BRCA1/2, (Jara et al, 2002a, Jara et al, 2002b, Gallardo et al 2004, Jara et al 2004, Jara et al 2006, Gallardo et al 2006, Gonzalez-Hormazabal et al 2011, Sánchez et al 2011, Alvarez et al 2017; Adaniel et al 2019). Eight other variants are reported: RAD51 (Jara et al 2007; Jara et al 2010), CHECK2 (González-Hormazabal et al 2008; González-Hormazabal et al 2010); ATM (González-Hormazabal et al 2008; Tapia et al 2008); BARD (González-Hormazabal et al 2012); TOX3 (Elematore et al 2014), FGFR2, MAP3K1 (Jara et al 2013) and PALB2 (Leyton et al 2015).

In Mexico, we found 18 studies since 2002, ten of them related to BRCA1/2, (Ruiz-Flores et al, 2002; Calderon-Garciduenas et al., 2005; Vidal-Millán et al. 2009; Vaca-Paniagua et al 2012; Nahleh et al 2015, Torres-Mejía et al 2015; Villarreal-Garza et al. 2015a; Villarreal-Garza et al 2015b; Fernández-López et al 2019; Zayas-Villanueva et al 2019. Nine other variants are reported: ATM (Calderon-Zuniga et al. 2014); FGFR2 (Murillo-Zamora et al. 2013); ERCC1/2(Gómez-Díaz et al. 2015); GSTM1, GSTT1, GSTP1, and GSTM3 (Jaramillo-Rangel et

al 2015, Soto-Quintana et al 2015); XRCC1 (Macías-Gómez et al 2015) and TP53 (Gallardo-Alvarado et al 2019). Quezada Urban et al. (2018) conducted a comprehensive study of germinal variants.

In Colombia, twelve studies were found from 2007, eleven of them studied the VP BRCA1/2 (Torres et al. 2007; Torres et al. 2009; Sanabria et al. 2009; Rodriguez et al. 2012; Hernández et al. 2014; Torres et al. 2017; Briceño-Balcázar et al 2017; Llinás-Quintero et al 2019; Vargas et al 2019; Cifuentes et al 2019; Cortés et al 2019). Cock-Rada et al conducted a multigene panel study.

Bahamas reports 5 studies as of 2011 (Donenberg et al. 2011; Akbari et al. 2014; Trottier et al. 2016; Bagherzadeh et al. 2020; George et al. 2021), report three genes with VP: BRCA1 (6.2-25%), BRCA2 (0.6-4%), and RAD51C (1.29%)

Argentina has reported since 2012, four studies (Solano et al. 2012; Solano et al. 2016; Solano et al. 2018; Cerratini et al. 2019), report four genes with VP: BRCA1 (1.8-16%), BRCA2 (4.5-7.87%), PALB2 (3.6%) and other genes (0.9%).

Two countries report three publications: in Puerto Rico, as of 2012 (Dutil et al. 2012; Diaz-Zabala et al. 2018; Dutil et al. 2019), find genes with VP in BRCA1 (0-8.7%), BRCA2 (0-39.1%) and other genes (8.32%), and Peru since 2015 (Abugattas et al 2015; Buleje et al 2015; Buleje et al 2017) reported two genes with VP in BRCA1 (4.13-16.6%) and BRCA2 (0.755-5.55%).

Four countries report two studies: in Uruguay (Delgado et al. 2011; Della et al. 2017) found four genes with VP: BRCA1 (4.7-6.6%), BRCA2 (8.9-11.9%), TP53 (3.7%) and another (2.22%); in Costa Rica (Gutiérrez et al. 2012; García et al 2012) reported two genes with VP in BRCA1 (0.86-0.9%) and BRCA2 (3.6-4.31%); in Trinidad and Tobago (Donenberg et al. 2016; George et al 2021) reported four genes with VP: BRCA1 (1.9-5.6%), BRCA2 (1.2-4.1%), PALB2 (0.7%), RAD51C and CHECK (0.2%); and in Jamaica (Lerner-Ellis et al 2017, George et al 2021) found five genes with VP: BRCA1 (0.19-0.55%), BRCA2 (0.19-1.11%), PALB2 (0.4-2.79%), NBN and STK11 (0.2%).

Nine countries have reported only one study: Cuba (Rodriguez et al 2008) found two genes with VP in BRCA2 (2.3%) and BRCA2 (0.3%); Venezuela (Lara et al 2012) reported two genes with VP in BRCA1 (10.3%) and BRCA2 (6.8%); in Nicaragua (Martínez et al 2021) they identified three genes with VP in BRCA2 (5%), TP53 (2.5%) and PALB2 (2.5%); in Guatemala (Ren et al 2021) they reported four genes with VP in BRCA1 (5.6%), BRCA2 (2.3%), TP53 (0.8%) and PALB2 (0.8%); in Barbados, Dominica, Haiti and the Cayman Islands (George et al 2021) reported three genes with VP: BRCA1 (7.6% in Barbados, 1.3% in Haiti and 0.1% in the Cayman Islands); BRCA2 (5.4% in Barbados, 4% in Haiti, 3.27% in Dominica and 3.2% in the Cayman

Islands); PALB2 (3.2% in Dominica and 1.3% in Haiti); Ecuador (López-Cortes et al 2015) analyzed the expression of the MTHFR gene.

The frequency of genes is related to VP for CaMaH

The most studied genes have been BRCA1/2, followed by CHECK, TP53, RAD51C, and other less frequent ones such as ATR ATM, MSH2, and PMS2. Table 2 shows the diversity of studies, usually descriptive studies, studying various population subgroups with a wide range of prevalence for the VP studied.

DISCUSSION

Since the last review conducted in 2019 by Urbina-Jara et al., 17 new studies were identified in just two years. The new studies carried out in the Caribbean (Barbados, Dominica, Haiti, Cayman Islands), and Central America (Nicaragua and Guatemala) reduce the research gap on CaMaH in developing countries, the socioeconomic limitations of these countries are the main cause of the lack of research on this topic, since genetic studies as stated by Chavarri-Guerra et al (2017), They are a highly specialized discipline that requires technological resources and trained personnel in the field.

As found in previous studies, the most analyzed genes are still BRCA 1/2, followed by TP53 and PALB2, but some countries have incorporated the analysis of other non-BRCA genes. There is heterogeneity in the frequency of VP in the 21 countries that have researched the subject, demonstrating the need to expand genetic screening in the population at risk.

The most frequent VP gene is BRCA2 (39.1%) research conducted in Puerto Rico, followed by BRCA1 (22.8%) reported in the Bahamas, Tp53 in third place with 8.6% research conducted in Brazil, and PALB2 with 3.6% in Argentina.

The results of this study reflect the existing inequality in CaMaH research in LAC and the need to foster research in lagging countries. This systematic review is the most recent to demonstrate scientific advances and remaining challenges, as well as being the first study to propose a classification based on knowledge gaps of CaMaH useful to promote research in less developed countries.

It is necessary to develop regional research policies on these issues, including cost-benefit studies to encourage the inclusion of genetic studies to at-risk populations and relatives of patients with CaMaH. It is also relevant to make specific efforts in regions, countries, and territories that have not studied CaMaH in their populations. It is considered that specific studies should be carried out in each country to improve the design of care programs for CaMa and CaMaH in particular, depending on the type of VP that is identified.

It is recommended that the existence, availability, and accessibility of genetic tests of diagnostic and therapeutic utility be studied, since it is a cornerstone to strengthen the approach, especially in less developed countries.

Regarding the limitations of the study, it was focused on the analysis of disparities in CaMaH research, rather than on the study of reported VPs and their trends.

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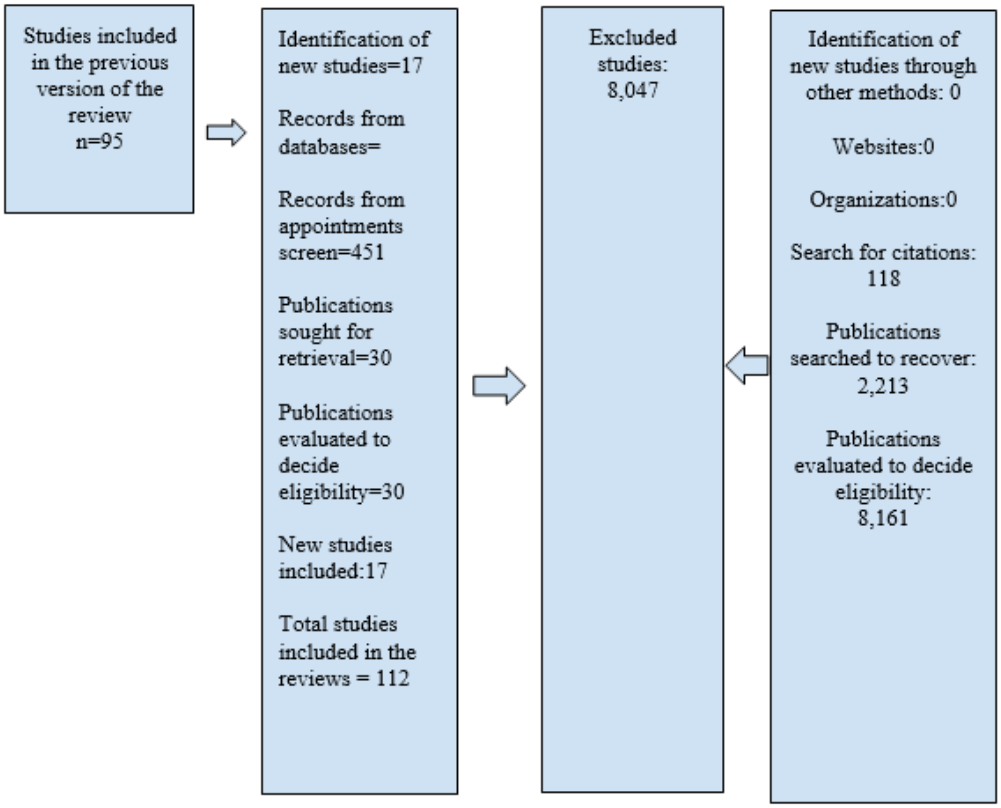
Conflict of interest:

The authors declare that they have no conflict of interest.

ANNEXES

Figure 1

Flowchart for study selection.



Source: PRISMA 2020 template.

Table 1

Frequency of research on CaMaH in LAC.

Classification	Countries	CaMa Studios (#)	Molecular studies CaMa #	Genetic studies on Bed Included in previous reviews	New genetic studies associated with CaMaH	Total, studies Genetics associated with CaMaH	F (%)
Group 1: Countries with genetic studies on CaMa	Brazil	3633	921	34	2	36	21/48 (44%)
	Colombia	371	125	9	3	12	
	Mexico	1920	580	16	2	18	
	Bahamas	18	10	2	3	5*	
	Argentina	734	243	3	1	4	
	Chile	491	148	19	1	20	
	Puerto Rico	204	58	2	1	3	
	Peru	217	61	3	0	3	
	Uruguay	101	26	2	0	2	
	Costa Rica	40	9	2	0	2	
	Trinidad and Tobago	61	1	0	2*	2*	
	Jamaica	46	8	0	2*	2*	
	Venezuela	59	13	1	0	1	
	Nicaragua	9	1	0	1	1	
	Guatemala	17	-	0	1	1	
	Dominica	22	2	0	1*	1*	
	Barbados	2	0	0	1*	1*	
	Haiti	5	2	0	1*	1*	
	Cayman Islands	0	0	0	1*	1*	
	Cuba	17	4	1	0	1	
	Ecuador	64	1	1	0	1	
	Subtotal	**7967**	**2213**	**95**	**17**	**112**	
Group 2: Countries with non-genetic studies on CaMa	Paraguay	38					10/48 (21%)
	Panama	25					
	Dominican Republic	17					
	Bolivia	15					
	El Salvador	12					
	Guyana	8					
	Suriname	5					
	Antigua and Barbuda	4					
	Honduras	4					
	Grenade	2					
	Subtotal	**194**					

Group 3: Countries without studies on CaMa	Belize Saint Kitts and Nevis Saint Vincent and the Grenadines Saint Lucia Aruba Guadeloupe Turks and Caicos Islands Virgin Islands Martinique Saint Barthélemy Anguilla Netherlands Antilles Bonaire Curaçao Bermuda Falkland Islands Monserrat						17/48 (35%)
Total		**8, 161**	**2, 213**	**95**	**17**	**112**	

*Caribbean countries included in the same study.

Source: Authors.

Table 2

Frequency of genes with VP in CaMaH in the last decade.

Countries	Author (year)	# sample	Type of I am a student	BRCA 1 (%)	BRCA 2 (%)	TP53 (%)	PALB2 (%)	Other genes (%)
Brazil	Carraro et al (2013)	54	D	13	7	2	-	0 CHECK
	Cury et al (2014)	28	CC	-	-	7.1	-	-
	Giacomazzi et al (2014)	59 815	D	-	-	3,4 8.6	-	-
	Felix et al (2014)	106	D	8.5	0	0.9 4	-	0 CHECK
	Fernández et al (2016)	349	D	14.04	7.45	-	-	-
	Palmero and Cabbage (2016)	18 40 7	D	0	5.5	0	-	14.3 CHECK
	German et al. (2017)	418	D	11.9	7.2	-	-	-
	Cyprian et al. (2019)	44	D	11.3	15.9	2.3	-	-
Colombia	Hernández et al (2014)	280	D	0.8	0.4	-	-	-
	Torres et al (2017)	68	D	1.4 5.5	8.8 1.5	-	-	-
	Briceño-Balcázar et al (2017)	256	D	8	4.4	-	-	-
	Cock-Rada et al (2018)	85	D	8.2	9.4	-	1.2	3.6 *ATM, MSH2 PMS2*
	Vargas et al (2019)	60	D	1.7	0	-	-	-
	Cifuentes et al (2019) (21)	58 20	CC	6.9	13.8	-	-	-
	Cuts and cabbage (2019)	104	D	17.3	14.4	-	-	-
Mexico	Cow-Paniagua and Cabbage (2012)	39	D	5.1	5.1	-	-	-
	Villarreal-Garza et al (2015)	96	D	Unspecified	Unspecified	-	-	-
	Torres-Mejía et al (2015)	810	D	2.5	1.8	-	-	-
	Villarreal-Garza, Weitzel et al (2015)	120	D	22.6	0.5	-	-	-
	Gallardo et al (2019)	78 509	CC	-	-	6.4	-	-
	Zayas-Villanueva et al (2019)	61 22 72	CC	11.9	1.9	-	-	-

Bahamas	Donenberg et al (2010)	214	D	22.8	0	-	-	-
	Akbari y col (2014)	214	D	23	4	-	-	-
	Trottier y col (2016)	705 1089	CC	0.09		-	-	-
	Bagherzadeh et al (2020)	387 653	CC	-	-	-	-	RAD51C 1.29
	George et al. (2021)	247	D	25.5	2.4	0.4	0	0
Argentina	Solano et al (2012)	134	D	17.2	11.2	-	-	-
	Solano et al (2016)	940	D	11.2	7.9	-	-	-
	Solano et al. (2018)	279	D	15.77	6.09	-	-	-
	Cerretini et al (2019)	112	D	1.8	4.5	-	3.6	0.9
Chile	González-Hormazabal et al (2011)	326	23	7.1		-	-	-
	Alvarez et al (2017)	453	D	7.06	8.61	-	-	-
	Daniel et al (2019)	315	D	8.2	8.8	-	1.3	CHECK2 0.9 RAD51C 0.6 CDH1 and RAD51D 0.3
Puerto Rico	Dutil y col (2012)	For example	D	8.7	39.1	-	-	-
	Díaz-Zabala et al (2018)	302	D	0	2.6	-	-	-
	Dutil y col (2019)	48	D	0	0	-	-	CHEK2 4.2 RAD51B 2.1 MUTYH 2.1
Peru	Abugattas et al (2015)	266	D	4.13	0.75	-	-	-
	Buleje et al (2017)	18 families	D	16.66	5.55	-	-	-
Uruguay	Delgado et al (2011)	42 families	D	4.8	11.9	-	-	-
	Della et al (2017)	135 families	D	6.7	8.9	3.7	-	RAD51C CHECK 2.22
Costa Rica	Gutierrez et al (2012)	111	D	0.9	3.6	-	-	-
	García-Jiménez et al (2012)	116	D	0.86	4.31	-	-	-

Trinidad and Tobago	Donenberg et al (2016)	268	D	5.6	4.1	-	0.7	-
	George et al. (2021)	298	D	6.4	4.03	0	0	0.2 RAD51C CHECK
Jamaica	Lerner-Ellis y col (2017)	179	D	0.55	1.11	-	2.79	-
	George et al. (2021)	183	D	1.1	1.1	0	2.2	NBN 1.1 STK11 1.1
Cuba	Rodriguez et al (2008)	99	D	0.3	2.3	-	-	-
Venezuela	Lara et al (2012)	58	D	10.3	6.8	-	-	-
Nicaragua	Martínez et al (2021)	39	D	-	5	2.5	2.5	-
Guatemala	Ren et al. (2021)	664	D	5.6	2.3	0.8	0.8	-
Barbados	George et al. (2021)	92	D	7.6	5.4	0	4.3	0
Dominica	George et al. (2021)	61	D	0	3.27	0	3.27	0
Haiti	George et al. (2021)	75	D	1.3	4	0	1.3	0
Cayman Islands	George et al. (2021)	62	D	1.6	3.2	0	0	1.6 ATM

Source: Authors.

NP: Not provided; D: Descriptive; CC: Case and Control

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