Table S1. Description of French Canadian study groups investigated in this study.

Study group[1]	Participant investigated	Number of participants (or families) investigated	Age range in years (average ± standard deviation)	Characteristics of study group and pathogenic *BRCA1* or *BRCA2* variant carrier status at selection for this study (FC ancestry[1])	Purpose of the study group (study phase)	Source of genetic information (number of participants subjected to genotyping or sequencing method) in this study[3]	Source of participants[1]
Familial OC cases	OC	20 (17)	24-74 (52.6±11.3)	Families with at least two epithelial OC, fallopian tube or primary peritoneal carcinomas within first-, second- or third-degree relatives; at least one OC case tested negative for pathogenic *BRCA1* and *BRCA2* variants by FC mutation-panel or clinical multi-gene panel testing.	Identify candidate variants in familial OC cases (phase I)	WES (20)	RRCancer biobank or adult hereditary cancer clinics [1–11]
Sporadic early-onset OC cases	OC	53	24-49 (44.1±5.5)	High-grade serous ovarian carcinoma cases diagnosed <50 years, not selected for family history of OC cancer; tested negative for pathogenic *BRCA1* and *BRCA2* variants by FC mutation-panel or clinical multi-gene panel testing.	Identify candidate variants in sporadic OC cases (phase I)	WES (53)	RRCancer biobank [1,2,4–8,10–16]
OC families	OC	49 (44)	31-77 (52.2±9.4)	Additional families with at least two OC cases diagnosed with epithelial OC, fallopian tube or primary peritoneal carcinomas within first-, second- or third-degree relatives within the same family branch; 71% of index OC cases tested positive for pathogenic *BRCA1* and *BRCA2* variants by FC mutation-panel or clinical multi-gene panel testing [5,7].	Determine carrier frequency of candidate variants in two OC case families (phase II)	WES (25) and TaqMan® genotyping (24)	RRCancer biobank [1–10,13,14]
HBOC families	OC or BC[2]	14 OC or 42 BC (56)	OC: 24-73 (49.6±13.7) BC: 30-61 (45.7±7.2)	Families with only one epithelial OC, fallopian tube or primary peritoneal carcinomas case and at least two invasive breast cancer cases (diagnosed <65 years) within first-, second- or third-degree relatives within the same family branch; 54% of index OC/BC cases tested positive for pathogenic *BRCA1* and *BRCA2* variants by FC mutation-panel or clinical multi-gene panel testing [5,7].	Determine carrier frequency of candidate variants in HBOC families (phase II)	WES (6) and TaqMan® genotyping (50)	RRCancer biobank [1,2,11,3–10]
Sporadic OC cases	OC	438	24-91 (60.8±10.5)	Epithelial OC, fallopian tube or primary peritoneal carcinomas cases (includes high-grade serous, high grade endometrioid, low-grade, ungraded serous or mixed subtypes tumours containing serous component were included) not selected for age at diagnosis or family history of OC cancer; 12% tested positive for pathogenic *BRCA1* and *BRCA2* variants by FC mutation-panel testing [14].	Determine carrier frequency of candidate variants in sporadic OC cases (phase II)	WES (52) and TaqMan® genotyping (386)	RRCancer biobank [3,14,17]

Sequencing-based controls[3]	Control	1025	Gen3G - newborn; MNI (not available); CARTaGENE - 41-70 (55.2±8.6)	Gen3G controls: 433 newborns (sex not divulged) from mothers were recruited between 2010-2013 to study genetics of glucose regulation in gestation and growth. MNI controls: 422 adult participants (202 females and 210 males) were recruited during different periods of time to study genetics of neurodegenerative or neurological disorders (311 affected; 11 unaffected and remaining unknown). CARTaGENE controls: 170 adult participants (52 females and 118 males) were recruited between 2009–2014 to study the causes of chronic diseases. Personal and family history of cancer, and pathogenic *BRCA1* and *BRCA2* variant carrier status unknown for all controls.	Determine carrier frequency of candidate variants in population-matched controls from DNA sequencing data (phase II)	WGS - Gen3G (433); WES - MNI (422) and CARTaGENE (170)	Gen3G project; MNI project; and CARTaGENE [17–23]
Genotyping-based controls[3]	Control	8493	39-71 (54.4±7.6)	CARTaGENE controls comprised of 4625 females and 3868 males recruited as described in sequencing-based controls: personal and family history of cancer, and pathogenic *BRCA1* and *BRCA2* variant carrier status unknown.	Determine carrier frequency of candidate variants in population-matched controls from genotyping-based data (phase II)	SNP array genotyping (100)	CARTaGENE [19–21]
Additional OC cases	OC	538	69% known: 24-89 (59.2±11.9)	Epithelial ovarian, fallopian tube or primary peritoneal carcinomas not categorized based on any defined inclusion criteria; pathogenic *BRCA1* and *BRCA2* variant carrier status unknown.	Identify additional carriers of candidate variants (phase III)	WES (44) and TaqMan® genotyping (494)	RRCancer biobank [24]

[1] All study groups contain participants recruited from the province of Quebec: familial ovarian cancer (OC) or invasive breast (BC) cases self-reported French Canadian (FC) ancestry [5,7]; the majority of sporadic OC (at least 88%) self-reported FC ancestry [14]; and additional OC cases self-reported FC ancestry by the Banque de tissus et données of the Réseau de recherche sur le cancer of the Fond de recherche du Québec – Santé (RRCancer biobank) or adult hereditary cancer clinics; Sherbrook University - glucose regulation in gestation and growth (Gen3G) controls included newborn whose mothers self-reported FC ancestry [18]; McGill University - Montreal Neurological Institute (MNI) controls were self-reported as FC ancestry [17,23]; and CARTaGENE defined FC status of controls based on being born in Quebec, having parents and all four grandparents born in Canada and having French as first language learned [19].

[2] BC case in closest familial relationship to OC was investigated where OC case was not available in the investigation of Hereditary Breast and Ovarian Cancer (HBOC) syndrome families study group.

[3] Genetic data derived from whole exome sequencing (WES) analyses or targeted TaqMan® genotyping assay for familial, sporadic or additional cases as described in Methods of this study; whole genome sequencing (WGS) analyses for Gen3G [18]; MNI [17]; and CARTaGENE [20–22] controls (sequencing-based controls); and three different single nucleotide (SNP) genotyping arrays analyses for CARTaGENE (genotyping-based controls): Affymetrix UK biobank Axiom® 2.0 gene chip (n=772) [19], Infinium® Global Screening Array (GSA) conducted in three phases (n=3159, 628, 3427) [19] and Infinium Omni 2.5 (n=510) [20,21].

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