COST Action BM1206

(30/04/2013 – 29/04/2017)

Cooperation Studies on Inherited Susceptibility to Colorectal Cancer

PROGRESS REPORT 2

(30/04/2013 – 21/12/2015)

This report is submitted by the MC Chair on behalf of the Management Committee and is validated by the Scientific Committee of the COST Association.

Confidentiality: the document will be made available to the public via the Action page on the COST website except for Section II.D.

Executive summary of the Progress Report:
(max.500 words) (to be completed by Action Chair describing the Action's progress with achieving the Action MoU objectives and generating outputs and impacts – see Annex 1 definitions)

Colorectal cancer (CRC) is one of the most frequent neoplasms worldwide and an important cause of morbidity in the developed world. It is necessary to identify individuals with a medium-high CRC risk in order to develop adequate preventive measures. The identification of inherited genetic factors involved in CRC susceptibility can help profile individual disease risk and may enable early screening and treatment monitoring.

COST Action BM1206 continued growing substantially including nowadays 141 participants from 44 research groups in 22 countries actively working on CRC genetics with 51.3% female participation, 55.8% ESR members and 26.6% inclusiveness. Six MC and WG joint meetings took place already, as well as four educational workshops, a training school and 8 STSMs (37.5% from Inclusiveness Target Countries). So far, 16 publications acknowledged COST support. Besides publications, dissemination tools included an initial brochure, an announcement at the European Journal of Human Genetics and a dedicated website (www.eucolongene.eu).

Achieving the Action MoU objectives

Up to 50 common, low-penetrance genetic variants are known nowadays for CRC genetic susceptibility. Since 2013, most of the newly identified variants have been discovered by participants of the present Action by performing genome-wide association studies (GWAS) and GWAS meta-analyses. Additional variants are precluded to be identified since some new GWAS will be finished soon. After its identification, functional meaning for these variants is generally lacking in most cases. Results were achieved by participants in this regard for some variants, as well as for their correlation with CRC survival exemplifying that they could act also as disease biomarkers. Additionally, new genes involved in high-penetrance genetic predisposition to CRC have been recently identified including *POLE*, *POLD1*, *BUB1*, *BUB3*, *FOCAD*, *GREM1*, *RPS20*, *NTHL1*, *FAN1* and *BLM*, and they were mainly identified by next generation sequencing (NGS) efforts performed by participants in this Action. Finally, the work initiated by participants regarding modelling low-penetrance variants and environmental factors together will permit to develop a high-risk profile for CRC in the near future.

Generating outputs and impacts

Besides achieving most MoU objectives, this Action helped to create expertise and capabilities among participants. This knowhow included GWAS and NGS design proficiency and data analysis pipelines that are available now in most groups. Moreover, competence in functional genomics increased with interaction between participants and availability of functional genomics platforms in some centres. Importantly, contact with another international CRC consortium (GECCO) was initiated recently for mutual benefit which will permit to generate additional results. Also, most participants agreed in the combination in a meta-analysis of NGS available data in the near future that will surely permit to identify additional genes involved in high-

penetrance genetic predisposition to CRC.

This Action is permitting to comprehensively understand the impact of inherited susceptibility to CRC and to help describing the genetic landscape of this disease, providing a highly innovative and unconditional tool for personalized CRC medicine with a future application in early screening and treatment monitoring for this disease. In this manner, these genetic factors will be used to decrease morbidity and mortality expectations for this neoplasm.

Summary assessment of Progress Review by Action Rapporteur:	
(max.500 words) (to be transferred by SO from Action Rapporteur report) SO enters Action Rapporteur 500 word summary here. SO enters Action Rapporteur	
Action Rapporteur	Name: Ursula GUNDERT-REMY Institution: Bundesinstitut fuer Risikobewertung, Thielallee 88-92, 14195 Berlin Country: Germany

Validation by Scientific Committee

This report was validated by the Scientific Committee on: <insert date of SC validation>

I. Progress Report

I.A. COST Action Profile

Objective/ Aim

Colorectal cancer (CRC) is one of the most frequent neoplasms worldwide and an important cause of morbidity in the developed world. It is necessary to identify individuals with a medium-high CRC risk in order to develop adequate preventive measures. The identification of inherited genetic factors involved in CRC susceptibility can help to profile individual disease risk and may enable early screening and treatment monitoring. Participants interested in networking in this Action include 19 research groups actively working on CRC genetics with high success and with access to more 50,000 CRC cases and 50,000 controls through cohorts in 11 countries. This Action will permit to articulate the cooperation between these research groups in Europe in order to comprehensively understand the impact of inherited susceptibility to CRC and to describe the genetic landscape of this disease, providing a highly innovative and unconditional tool for personalized CRC medicine with a future application in early screening and treatment monitoring for this disease.

Details

MoU:	11/01/2013	Start of Action:	30/04/2013
CSO approval date:	21/11/2012	End of Action:	29/4/2017

COST Member Countries and Cooperating State having accepted the MoU

Country	Date	Country	Date	Country	Date	Country	Date
Austria	07/01/2013	Croatia	06/03/2014	Cyprus	17/06/2013	Czech Republic	06/06/2013
Finland	17/12/2012	France	14/06/2013	Germany	17/01/2013	Greece	10/04/2014
Ireland	13/01/2014	Israel	29/05/2013	Italy	14/02/2013	Malta	12/03/2013
Netherlands	21/01/2013	Norway	22/10/2013	Portugal	11/01/2013	Romania	07/06/2013
Serbia	28/02/2014	Spain	28/11/2012	Sweden	07/02/2013	Turkey	07/05/2013
United Kingdom	28/11/2012	fYR Macedonia	27/02/2013				

Total: 22

Intentions to Accept the MoU

0”

Other participants:

Institution Name	Country
University of California, Davis (UC Davis)	United States of America

Contacts

Chair/ Vice Chair

Position	Name	Contact details	Country	Date of PhD:	Gender
Chair:	Sergi CASTELLVI-BEL	Institut dInvestigacions Biomdiques August Pi i Sunyer (IDIBAPS), Centre Esther Koplovitz (CEK), Gastrointestinal and Pancreatic Oncology, Rossello 153, Planta 4, 08036 Barcelona,Spain +34932275400 ext.4183 sbel@clinic.ub.es	ES	1996	M

Vice Chair:	Annika LINDBLOM	Karolinska Institutet, Solna, S17176, Stockholm, Sweden 46+8+51775248 annika.lindblom@ki.se	SE	1993	F

Working Group Leaders

WG #	WG Title	WG Leader	Country	Date of PhD:	Gender	Number of participants
1	Genetic Association Studies	Ian TOMLINSON	UK	1988	M	113/141
2	Functional Genomics	José Luis GÓMEZ-SKARMETA	ES	1995	M	66/141
3	Next Generation Sequencing	Tom van WEZEL	NL	1998	M	94/141
4	Statistical Modeling	Albert TENESA	UK	2003	M	113/141

Other positions if applicable (STSM Coordinator, WG Vice Leader, Task Force Leader…)

Position	Name	Country	Date of PhD:	Gender
STSM Coordinator	Kari HEMMINKI	DE	1973	M
STSM Committee	Andrea GSUR	AT	1994	F
STSM Committee	Lauri AALTONEN	FI	1994	M
WG1 Vice Leader	Dina RUANO	NL	2007	F
WG2 Vice Leader	Richard HOULSTON	UK	1992	M
WG3 Vice Leader	Claire PALLES	UK	2009	F
WG4 Vice Leader	Ceres FERNÁNDEZ-ROZADILLA	UK	2011	F
Financial rapporteur	Clara RUIZ-PONTE	ES	1996	F
Financial rapporteur	Jochen HAMPE	DE	1996	M
Publications Committee	Ian TOMLINSON	UK	1988	M
Publications Committee	Vanessa PETRONI	MT	--	F
Website Committee	Sergi CASTELLVI-BEL	ES	1996	M
Website Committee	Pavel VODICKA	CZ	1986	M

Action website:	www.eucolongene.eu

I.B. Progress with MoU objectives and deliverables and additional outputs

MoU objectives

MoU objective	Achieved Yes/ Partially/ No	Evidence of (partial) achievement including hyperlink to enable assessment of the achievement[1]. Justification if full achievement is not forseen
Identification of new CRC susceptibility variants	Partially	Up to 50 common, low-penetrance variants are known nowadays for CRC genetic susceptibility. Since 2013, most of the newly identified variants have been tackled by participants, and some of them acknowledged the present Action as exemplified by the following links to published articles: http://www.ncbi.nlm.nih.gov/pubmed/24743384 http://www.ncbi.nlm.nih.gov/pubmed/24465592 http://www.ncbi.nlm.nih.gov/pubmed/24218287 http://www.ncbi.nlm.nih.gov/pubmed/24737748 http://www.ncbi.nlm.nih.gov/pubmed/24978480 http://www.ncbi.nlm.nih.gov/pubmed/26553438 http://www.ncbi.nlm.nih.gov/pubmed/26621817
Functional links for CRC susceptibility variants	Partially	Functional meaning for CRC genetic susceptibility variants is lacking in most cases after its identification by genetic association studies. Some participants worked on this objective and one related published article acknowledged the present Action: http://www.ncbi.nlm.nih.gov/pubmed/25131200 It should be noted that some of the functional work has been also performed as part of the "New predisposition genes for CRC in families with unknown genetic basis" objective since it is nowadays necessary to prove the functional effect of a candidate genetic variant in order to corroborate its involvement in disease pathogenesis.
Genetic variants involved in CRC survival and treatment toxicity	Yes	CRC genetic susceptibility variants could also act as biomarkers for CRC survival and treatment response. Some common, low-penetrance genetic variants were tested as biomarkers for CRC survival. Two related published articles acknowledged the present Action: http://www.ncbi.nlm.nih.gov/pubmed/23712746 http://www.ncbi.nlm.nih.gov/pubmed/25350395
Genetic variants enriched in CRC subgroups	No	This objective was investigated in the past but not further pursued by participants.The main reason for abandoning it was that dividing cohorts by different subgroups reduces statistical power to solidly detect genetic associations. The identification of new variants was prioritized as shown in the first objective.
New predisposition genes for CRC in families with unknown genetic basis	Partially	New genes involved in genetic predisposition to CRC have been mainly identified recently by participants in this Action through next generation sequencing (NGS). Some of them have acknowledged the present Action as exemplified by the following links to

1 The links to the outputs and deliverables will be used by the Action Rapporteur in assessing the progress.

		published articles: http://www.ncbi.nlm.nih.gov/pubmed/23837913 http://www.ncbi.nlm.nih.gov/pubmed/24587672 http://www.ncbi.nlm.nih.gov/pubmed/25370038 http://www.ncbi.nlm.nih.gov/pubmed/25058500 http://www.ncbi.nlm.nih.gov/pubmed/25860647
Interactions between CRC susceptibility variants and environment	Partially	CRC genetic susceptibility variants may also act together with environmental risk factors for this disease. Data available from different cohorts regarding CRC genetic susceptibility variants and environmental factors is being gathered in a database. Those groups that accept to combine their data will enable this objective to be achieved.
High risk CRC profile of inherited variants by statistical modelling	Partially	Likewise, the previously mentioned data will permit to develop a high-risk CRC profile using data from cohorts that accept to collaborate.

MoU deliverables

MoU deliverable	Level of progress[1]	Evidence of (partial) delivery achievement including hyperlink to enable assessment of the delivery[1]. Justification if full achievement is not forseen
Excel file for WG1 and WG4 from CRC cohorts	Completed	This file summarized all genotypic, phenotypic and environmental data available from the differents CRC cohorts involved in this Action.
WG1 and WG4 database	Ongoing	This database will gather previously mentioned data from tose CRC cohorts that agreed to combine it for WG1 and especially for WG4.
Genome-wide association study in Ankara, Turkey (WG1/WG4)	Ongoing	Genome-wide association studies (GWAS) are being performed using a family-based approach (51 trios of CRC cases and healthy parents) and a population-based approach (1,000 cases and 1,000 controls)
Genome-wide association study in Zagreb, Croatia (WG1/WG4)	Ongoing	Genome-wide association study (GWAS) is being performed using a population-based approach (cases and controls recruitment finishing)
Genome-wide association study in Vienna, Austria (WG1/WG4)	Ongoing	Genome-wide association study (GWAS) is being wrapped up using a population-based approach (978 cases, 636 high-risk polyps and 4,294 controls)
Genome-wide association study in Leiden, Netherlands (WG1/WG4)	Ongoing	Genome-wide association study (GWAS) is being performed for colorectal polyposis (400 cases and 3,000 controls)
Functional Genomics platform in Sevilla, Spain (WG2)	Completed	https://www.upo.es/CABD/AquaticVertebratesPlatform/ Our Action has permitted to open up this functional genomics platform directed by the WG2 leader to the rest of participants in our Action. One of the STSM took place in this platform and the training school in GP4 will also be organized here.

Functional Genomics platform in Belgrade, Serbia (WG2)	Completed	http://www.imgge.bg.ac.rs/index.php/en/facilities/zebra Our Action enabled also to make this Zebra fish Unit available to the rest of participants interested in functional genomics studies using this model.
Functional Genomics platform in Istanbul, Turkey (WG2)	Completed	Expertise in functional studies including migration assays, xenograft models and genome editing is available to interested participants.
Colonomics study in Barcelona (IDIBELL), Spain (WG2)	Completed	Data in germline and somatic CRC genomics is being used to identify functional variants behind common, low-penetrance loci as shown in the following articles: http://www.ncbi.nlm.nih.gov/pubmed/24760461
Functional studies platform in *Saccharomyces cerevisiae* and *Schizosaccharomyces pombe* (WG2) in Oxford, UK	Completed	This platform permits to evaluate the functionality of *POLE*/*POLD1* variants. This platform was first reported in the following article: http://www.ncbi.nlm.nih.gov/pubmed/23263490
Functional studies platform in *Schizosaccharomyces pombe* (WG2) in Barcelona (IDIBAPS), Spain	Ongoing	This platform permits to evaluate the functionality of *POLE*/*POLD1* variants. This platform has not been reported yet.
Next generation sequencing analysis pipeline (WG3) in Oxford, UK	Completed	As part of the efforts directed to identify new predisposition genes for CRC, a NGS pipeline was generated by this group. This pipeline was used to identify *POLE* and *POLD1* as shown in the following article: http://www.ncbi.nlm.nih.gov/pubmed/23263490
Next generation sequencing analysis pipeline (WG3) in London, UK	Completed	As part of the efforts directed to identify new predisposition genes for CRC, a NGS pipeline was generated by this group. This pipeline was used to analyse exome data regarding known CRC genes as shown in the following article: http://www.ncbi.nlm.nih.gov/pubmed/25559809
Next generation sequencing analysis pipeline (WG3) in Helsinki, Finlnad	Completed	As part of the efforts directed to identify new predisposition genes for CRC, a NGS pipeline was generated by this group. This pipeline was used to identify 11 candidate genes for CRC predisposition as shown in the following article: http://www.ncbi.nlm.nih.gov/pubmed/24146633
Next generation sequencing analysis pipeline (WG3) in Barcelona (IDIBAPS), Spain	Completed	As part of the efforts directed to identify new predisposition genes for CRC, a NGS pipeline was generated by this group. This pipeline was used to identify 6 candidate genes for CRC predisposition as shown in the following article: http://www.ncbi.nlm.nih.gov/pubmed/25058500
Next generation sequencing analysis pipeline (WG3) in Trondheim, Norway	Completed	As part of the efforts directed to identify new predisposition genes for CRC, a NGS pipeline was generated by this group. This pipeline was used to

		identify a new mutation in *POLE* as shown in the following article: http://www.ncbi.nlm.nih.gov/pubmed/25860647
Next generation sequencing analysis pipeline (WG3) in Nijmegen, Netherlands	Completed	As part of the efforts directed to identify new predisposition genes for CRC, a NGS pipeline was generated by this group. This pipeline was used to identify *BUB1*, *BUB3*, *FOCAD*, *NTHL1*, *BLM* and as well as several other candidate genes as shown in the following articles: http://www.ncbi.nlm.nih.gov/pubmed/23747338 http://www.ncbi.nlm.nih.gov/pubmed/25712196 http://www.ncbi.nlm.nih.gov/pubmed/25892863 http://www.ncbi.nlm.nih.gov/pubmed/25938944 http://www.ncbi.nlm.nih.gov/pubmed/26358404
Next generation sequencing analysis pipeline (WG3) in Barcelona (IDIBELL), Spain	Completed	As part of the efforts directed to identify new predisposition genes for CRC, a NGS pipeline was generated by this group. This pipeline was used to identify *FAN1* as shown in the following article: http://www.ncbi.nlm.nih.gov/pubmed/26052075
Next generation sequencing analysis pipeline (WG3) in Leiden, Netherlands	Completed	As part of the efforts directed to identify new predisposition genes for CRC, a targeted NGS pipeline was generated by this group for *MLH1*, *MSH2*, *MSH6*, *PMS2*, *POLE* and *POLD1*. This pipeline was used in the following article: http://www.ncbi.nlm.nih.gov/pubmed/26648449
Next generation sequencing analysis pipeline (WG3) in Leiden, Netherlands	Ongoing	A NGS pipeline has been generated to identify new predisposition genes for CRC. This pipeline is not yet reported.
Next generation sequencing analysis pipeline (WG3) in Skopje, Macedonia	Ongoing	A NGS pipeline has been generated to identify new predisposition genes for CRC. This pipeline is not yet reported.
Next generation sequencing analysis pipeline (WG3) in Madrid, Spain	Ongoing	A NGS pipeline has been generated to identify new predisposition genes for CRC. This pipeline is not yet reported.
Next generation sequencing analysis pipeline (WG3) in Santiago, Spain	Ongoing	A NGS pipeline has been generated to identify new predisposition genes for CRC. This pipeline is not yet reported.
Next generation sequencing analysis pipeline (WG3) in Edinburgh, UK	Ongoing	A NGS pipeline has been generated to identify new predisposition genes for CRC. This pipeline is not yet reported.

Co-authored publications and FP7/ H2020 proposals
The co-authored publications and FP7/ H2020 proposals/ projects resulting from the Action are listed on the page following the “Additional outputs and achievements” section

Additional outputs and achievements

Please describe any other outputs and achievements that have resulted or are in progress, focusing in particular on those that contribute to the COST mission of "COST enables break-through scientific developments leading to new concepts and products and thereby contributes to strengthen Europe's research and innovation capacities."

Regarding WG3 that corresponds to Next Generation Sequencing (NGS) efforts to identify new predisposition genes for CRC, most participants agreed in the combination in a **meta-analysis of NGS available data** in the near future. It should be noted that there is still some reluctance to perform this combination. The reason behind it is that most groups are still in the process of getting individual results published and they want to wait until then. On the other hand, a look-up service was indeed agreed by some groups (Leiden, Nijmegen, Madrid, Barcelona, Santiago) which can be used to check if variants in specific genes are present in external NGS datasets in order to help prioritize them.

Regarding the **relationship with other CRC consortia**, two agreements were pursued outside our Action including GECCO and EPIC groups. The Genetics and Epidemiology of Colorectal Cancer Consortium (GECCO, http://research.fhcrc.org/peters/en/genetics-and-epidemiology-of-colorectal-cancer-consortium.html) is a collaborative effort of researchers from North America, Australia, and Europe, using data from over 40,000 participants. The coordinating centre for this international consortium is based at the Fred Hutchinson Cancer Research Centre (Principal investigator: Ulrike Peters). There was a first meeting in October 2015 during the American Society of Human Genetics meeting in Baltimore between Ulrike Peters, Tabitha Harrison, representing GECCO, and Ian Tomlinson (WG1 leader) and Sergi Castellvi-Bel (Chair of the Action) representing our consortium. Several topics were discussed including GWAS and NGS data combination, and authorship policy. As a first joint task, a meta-analysis of the exome array data available in both consortia will be performed. Our exome array data was recently published (http://www.ncbi.nlm.nih.gov/pubmed/26553438). Other joint tasks will be pursued in the near future for mutual benefit.

Whereas collaboration with GECCO can be considered successful, the attempts to contact in 2014 the part of EPIC consortium (European Prospective Investigation into Cancer and Nutrition, http://epic.iarc.fr/) dedicated to CRC (Elio Riboli, Paolo Vineis) simply yielded no answer.

Regarding **interactions with other Actions**, they took place so far to some extent with COST Actions BM1006, BM1106 and BM1204.

Co-authored publications and FP7/ H2020 proposals

Co-authored publications

Enter in the table below only publications on the topic of the Action, co-authored by at least two Action participants from two different countries participating in the Action and for which the Action networking added value. A maximum of ten publications may be entered. If the Action has more than ten such publications the Core Group should select the ten most significant ones to include in the table below.

NO.	Bibliographic data (including: Title, Authors, Title of the periodical or the series, Issue number or volume, Publisher, Year of publication, Relevant pages)	Main author	Number of authors	Ac	WGs involved in publication	Date of submission (must be after Action start date)	Expected date of publication (if not already published)	Persistent link to publicly available version of the paper (if available) or the abstract	Is/Will open access[3] provided to this publication?	Is/ will COST be cited/ acknowledged in the publication?	Are/ will COST funds (be) implicated in this publication	Relevance to H2020 Societal Challenges[4]?	Is it peer-reviewed?	Was the added value of the Action Networking necessary for the publication	Impact Factor (if applicable)
1	Fernandez-Rozadilla C, Cazier JB, Tomlinson I, Brea-Fernández A, Lamas MJ, Baiget M, López-Fernández LA, Clofent J, Bujanda L, Gonzalez D, de Castro L; EPICOLON Consortium, Hemminki K, Bessa X, Andreu M, Jover R, Xicola R, Llor X, Moreno V, Castells A, Castellví-Bel S, Carracedo A, Ruiz-Ponte C. A genome-wide association study on copy-number variation identifies a 11q11 loss as a candidate susceptibility variant for colorectal cancer. Hum Genet. 2014 May;133(5):525-34.	Clara Ruiz-Ponte	23	Pe	WG1	29/08/2013	Published	http://www.ncbi.nlm.nih.gov/pubmed/24218287	no	yes	no	yes “Health, demographic change and wellbeing”	yes	yes	4.824
2	Pardini B, Verderio P, Pizzamiglio S, Nici C, Maiorana MV, Naccarati A, Vodickova L, Vymetalkova V, Veneroni S, Daidone MG, Ravagnani F, Bianchi T, Bujanda L, Carracedo A, Castells A, Ruiz-Ponte C, Morreau H, Howarth K, Jones A, Castellví-Bel S, Li L, Tomlinson I, Van Wezel T, Vodicka P, Radice P, Peterlongo P; EPICOLON Consortium. Association between CASP8 -652 6N del polymorphism (rs3834129) and colorectal cancer risk: results from a multi-centric study. PLoS One. 2014 Jan 21;9(1):e85538.	Paolo Peterlongo	27	Pa	WG1	27/09/2013	Published	http://www.ncbi.nlm.nih.gov/pubmed/24465592	yes	yes	yes	yes “Health, demographic change and wellbeing”	yes	yes	3.234
3	Esteban-Jurado C, Garre P, Vila M, Lozano JJ, Pristoupilova A, Beltrán S, Abulí A, Muñoz J, Balaguer F, Ocaña T, Castells A, Piqué JM, Carracedo A, Ruiz-Ponte C, Bessa X, Andreu M, Bujanda L, Caldés T, Castellví-Bel S. New genes emerging for colorectal cancer predisposition. World J Gastroenterol. 2014 Feb 28;20(8):1961-71.	Sergi Castellvi-Bel	19	Es	WG3	01/10/2013	Published	http://www.ncbi.nlm.nih.gov/pubmed/24587672	yes	yes	no	yes “Health, demographic change and wellbeing”	yes	yes	2.369
4	Abulí A, Bujanda L, Muñoz J, Buch S, Schafmayer C, Valeria Maiorana M, Veneroni S, van Wezel T, Liu T, Westers H, Esteban-Jurado C, Ocaña T, Piqué JM, Andreu M, Jover R, Carracedo A, Xicola RM, Llor X, Castells A; EPICOLON Consortium, Dunlop M, Hofstra R, Lindblom A, Wijnen J, Peterlongo P, Hampe J, Ruiz-Ponte C, Castellví-Bel S. The MLH1 c.1852_1853delinsGC (p.K618A) variant in colorectal cancer: genetic association study in 18,723 individuals. PLoS One. 2014 Apr 17;9(4):e95022.	Sergi Castellvi-Bel	28	Bu	WG1	16/12/2013	Published	http://www.ncbi.nlm.nih.gov/pubmed/24743384	yes	yes	yes	yes “Health, demographic change and wellbeing”	yes	yes	3.234
5	Whiffin N, Hosking FJ, Farrington SM, Palles C, Dobbins SE, Zgaga L, Lloyd A, Kinnersley B, Gorman M, Tenesa A, Broderick P, Wang Y, Barclay E, Hayward C, Martin L, Buchanan DD, Win AK, Hopper J, Jenkins M, Lindor NM, Newcomb PA, Gallinger S, Conti D, Schumacher F, Casey G, Liu T; Swedish Low-Risk Colorectal Cancer Study Group, Campbell H, Lindblom A, Houlston RS, Tomlinson IP, Dunlop MG. Identification of susceptibility loci for colorectal cancer in a genome-wide meta-analysis. Hum Mol Genet. 2014 Sep 1;23(17):4729-37.	Richard Houlston, Ian Tomlinson, Malcolm Dunlop	32	Wh	WG1	07/01/2014	Published	http://www.ncbi.nlm.nih.gov/pubmed/24737748	no	yes	no	yes “Health, demographic change and wellbeing”	yes	yes	6.393
6	Lu S, Pardini B, Cheng B, Naccarati A, Huhn S, Vymetalkova V, Vodickova L, Buchler T, Hemminki K,	Asta Försti	11	Vy	WG1	03/06/2014	Published	http://www.ncbi.nlm.nih.gov/pub	yes	yes	no	yes “Health,	ye	yes	3.234

3 Open Access is defined as free of charge access for anyone via Internet. Please answer "yes" if the open access to the publication is already established and also if the embargo period for open access is not yet over but you intend to establish open access afterwards.

4 H2020 Societal Challenges are “Health, demographic change and wellbeing”; “Food security, sustainable agriculture and forestry, marine and maritime and inland water research, and the Bioeconomy”; “Secure, clean and efficient energy”; “Smart, green and integrated transport”; “Climate action, environment, resource efficiency and raw materials”; “Europe in a changing world - inclusive, innovative and reflective societies”; “Secure societies - protecting freedom and security of Europe and its citizens”

	Vodicka P, Försti A. Single nucleotide polymorphisms within interferon signaling pathway genes are associated with colorectal càncer susceptibility and survival. PLoS One. 2014 Oct 28;9(10):e111061.							med/25350395			demographic change and wellbeing”	s		
7	Lewis A, Freeman-Mills L, de la Calle-Mustienes E, Giráldez-Pérez RM, Davis H, Jaeger E, Becker M, Hubner NC, Nguyen LN, Zeron-Medina J, Bond G, Stunnenberg HG, Carvajal JJ, Gomez-Skarmeta JL, Leedham S, Tomlinson I. A polymorphic enhancer near GREM1 influences bowel cancer risk through differential CDX2 and TCF7L2 binding. Cell Rep. 2014 Aug 21;8(4):983-90.	Ian Tomlinson	16	Ca	WG2	04/04/2014	Published	http://www.ncbi.nlm.nih.gov/pubmed/25131200	yes	yes	no	yes “Health, demographic change and wellbeing”	yes	yes	8.358
8	Esteban-Jurado C, Vila-Casadesús M, Garre P, Lozano JJ, Pristoupilova A, Beltran S, Muñoz J, Ocaña T, Balaguer F, López-Cerón M, Cuatrecasas M, Franch-Expósito S, Piqué JM, Castells A, Carracedo A, Ruiz-Ponte C, Abulí A, Bessa X, Andreu M, Bujanda L, Caldés T, Castellví-Bel S. Whole-exome sequencing identifies rare pathogenic variants in new predisposition genes for familial colorectal cancer. Genet Med. 2015 Feb;17(2):131-42.	Sergi Castellvi-Bel	22	Es	WG3	08/04/2014	Published	http://www.ncbi.nlm.nih.gov/pubmed/25058500	yes	yes	no	yes “Health, demographic change and wellbeing”	yes	yes	7.329
9	Timofeeva MN, Kinnersley B, Farrington SM, Whiffin N, Palles C, Svinti V, Lloyd A, Gorman M, Ooi LY, Hosking F, Barclay E, Zgaga L, Dobbins S, Martin L, Theodoratou E, Broderick P, Tenesa A, Smillie C, Grimes G, Hayward C, Campbell A, Porteous D, Deary IJ, Harris SE, Northwood EL, Barrett JH, Smith G, Wolf R, Forman D, Morreau H, Ruano D, Tops C, Wijnen J, Schrumpf M, Boot A, Vasen HF, Hes FJ, van Wezel T, Franke A, Lieb W, Schafmayer C, Hampe J, Buch S, Propping P, Hemminki K, Försti A, Westers H, Hofstra R, Pinheiro M, Pinto C, Teixeira M, Ruiz-Ponte C, Fernández-Rozadilla C, Carracedo A, Castells A, Castellví-Bel S, Campbell H, Bishop DT, Tomlinson IP, Dunlop MG, Houlston RS. Recurrent Coding Sequence Variation Explains Only A Small Fraction of the Genetic Architecture of Colorectal Cancer. Sci Rep. 2015 Nov 10;5:16286.	Malcolm Dunlop	61	Ti	WG1	20/03/2015	Published	http://www.ncbi.nlm.nih.gov/pubmed/26553438	yes	yes	yes	yes “Health, demographic change and wellbeing”	yes	yes	5.578
10	Cheng TH, Thompson D, Painter J, O'Mara T, Gorman M, Martin L, Palles C, Jones A, Buchanan DD, Ko Win A, Hopper J, Jenkins M, Lindor NM, Newcomb PA, Gallinger S, Conti D, Schumacher F, Casey G, Giles GG, Pharoah P, Peto J, Cox A, Swerdlow A, Couch F, Cunningham JM, Goode EL, Winham SJ, Lambrechts D, Fasching P, Burwinkel B, Brenner H, Brauch H, Chang-Claude J, Salvesen HB, Kristensen V, Darabi H, Li J, Liu T, Lindblom A, Hall P, de Polanco ME, Sans M, Carracedo A, Castellvi-Bel S, Rojas-Martinez A, Aguiar Jnr S, Teixeira MR, Dunning AM, Dennis J, Otton G, Proietto T, Holliday E, Attia J, Ashton K, Scott RJ, McEvoy M, Dowdy SC, Fridley BL, Werner HM, Trovik J, Njolstad TS, Tham E, Mints M, Runnebaum I, Hillemanns P, Dörk T, Amant F, Schrauwen S, Hein A, Beckmann MW, Ekici A, Czene K, Meindl A, Bolla MK, Michailidou K, Tyrer JP, Wang Q, Ahmed S, Healey CS, Shah M, Annibali D, Depreeuw J, Al-Tassan NA, Harris R, Meyer BF, Whiffin N, Hosking FJ, Kinnersley B, Farrington SM, Timofeeva M, Tenesa A, Campbell H, Haile RW, Hodgson S, Carvajal-Carmona L, Cheadle JP, Easton D, Dunlop M, Houlston R, Spurdle A, Tomlinson I. Meta-analysis of genome-wide association studies identifies common susceptibility polymorphisms for colorectal and endometrial cancer near SH2B3 and TSHZ1. Sci Rep. 2015 Dec 1;5:17369.	Ian Tomlinson	101	Pa	WG1	20/03/2015	Published	http://www.ncbi.nlm.nih.gov/pubmed/26621817	yes	yes	no	yes “Health, demographic change and wellbeing”	yes	yes	5.578

FP7/ H2020 Proposals and projects

This table contains FP7/ H2020 proposals/ projects spinning off from Action activities and including in the proposing consortium at least three Action participants from at least three different countries participating in the Action.

NO.	Title	Name and country of main proposer	Number of proposers	Action participants listed among the proposers (Name, country, role[3] in the Action)	Date submitted	Date results expected	Call identifier	Relevance to H2020 Societal Challenges[4]?	Was the added value of the Action Networking necessary for the proposal / project?
Projects									
1	Transcan project “Familial cancer” April 2014-April 2016	Kari Hemminki, DE	3	Rolf Sijmons, NL, COST Action participant	24/06/2013	October 2013		yes “Health, demographic change and wellbeing”	yes
2									
Proposals									
	USE OF GENOMIC DATA TO PREVENT COLORECTAL CANCER IN THE GENERAL POPULATION (STRATPREVCOLOCAN)	UK	10	Ian Tomlinson, UK, WG1 leader Lauri Aaltonen, FI, STSM committee Sergi Castellvi-Bel, ES, Chair of the Action Clara Ruiz-Ponte, ES, Financial Rapporteur Hilal Ozdag, TR, Participant Richard Houlston, UK, WG2 vice leader Tom van Wezel, NL, WG3 leader	20/08/2014	13/01/2015	H2020-PHC-2014-two-stage	yes “Health, demographic change and wellbeing”	yes

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I.C. Networking

Added value of the Networking

Please describe here the added value of the networking, highlighting in particular anything that would not have happened without the Action networking.

The present Action has enabled so far research interactions between 151 participants working on CRC genetic predisposition from 44 research groups in 22 countries. Their expertise is diverse ranging from clinical, genetics and molecular aspects related to this neoplasm. Participants' research could be encompassed within one or more of the established working groups (WG): Genetic association studies (WG1), Functional studies (WG2), Next generation sequencing (WG·) and Statistical modeling (WG4).

The Action organized 2 annual meetings that included a previous workshop dedicated to one of the WG and a subsequent joint Management Committee and Working Groups meeting to discuss further aspects of own research.

WG1 Genetic association studies: networking facilitated the identification of additional common, low-penetrance genetic variants involved in genetic susceptibility to CRC. This goal was achieved by performing combination or meta-analysis of GWAS sets from the participating cohorts and it was permitted by the ample sample size that boosted statistical power. Besides, several GWAS sets are in progress which precludes the identification of additional common, low-penetrance genetic variants and new meta-analysis in the near future. Additionally, CRC genetic susceptibility variants could also act as biomarkers for CRC survival and treatment response and some of them were tested as biomarkers for CRC survival. Finally, collaboration has been set recently with another international CRC consortium (GECCO) that will permit to know better the genetic predisposition landscape for this disease by identifying additional genetic variants.

WG2 Functional genomics: networking permitted to connect geneticists with molecular biologists with more expertise in functional genomics. This field is necessary to find the functional link behind common, low-penetrance genetic variants involved in genetic susceptibility to CRC, and also to prove that a genetic variant identified by next generation sequencing studies is pathogenic by altering gene function and being involved in CRC occurrence. Several participants with more expertise in WG2 have available functional genomics platforms in their groups that were made available to other participants in this Action, and in some cases allowed the successful identification of the underneath functional link.

WG3 Next generation sequencing (NGS): networking allowed the interaction of the different research groups working in this field. Focused CRC cohorts and different strategies were put in common in meetings. Our first training school was devoted to this WG. Several new genes involved in high-penetrance CRC genetic predisposition were identified by members of this consortium. Participants working on this WG developed NGS analysis pipelines Combination of the different datasets generated was also discussed and so far a look-up service permitted data sharing between some groups. A meta-analysis of most datasets is expected in the near future after individual results from different groups are published.

WG4 Statistical modeling: networking put together several CRC cohorts including all associated genotypic, phenotypic and environmental data. Available data is gathered in an Excel file and it will permit to generate a database including data to be combined from agreeing cohorts.

Extent of the networking

Describe the extent of the networking among the participants in the Action. Were all participants integrated into the networking equally? Were those targeted by COST policies on Inclusiveness Target Countries (ITCs), Early Career Investigators (ECIs)/ Young Researchers, and gender balance fully integrated into the Action networking?

The present Action was based on a previous consortium. Those participants already included in it were more integrated in research activities and interactions. However, most participants have reached nowadays an acceptable level of integration.

Regarding COST policies, participants from Inclusiveness Target Countries corresponded to 26.6%, early

career investigators were 55.8% and gender balance was achieved among participants (51.3% females)

I.D. Impacts

The impacts that have resulted, or might result from the Action are described in the following table.

Description of the impact	Type of impact[5]	Timing of impact[6]
Identification of new CRC susceptibility variants by GWAS	Scientific/ technological	Achieved
Large scale meta-analysis of existing CRC GWAS	Scientific/ technological	Achieved
Identification of additional new CRC susceptibility variants by GWAS	Scientific/ technological	Foreseen within 2 years
Additional large scale meta-analysis of existing CRC GWAS	Scientific/ technological	Foreseen within 2 years
Functional links for CRC susceptibility variants	Scientific/ technological	Achieved
Functional links for additional CRC susceptibility variants	Scientific/ technological	Foreseen within 2 years
Genetic variants involved in CRC survival and treatment toxicity	Scientific/ technological	Achieved
Genetic variants involved in CRC survival and treatment toxicity	Scientific/ technological	Foreseen within 2 years
Identification of new predisposition genes for CRC in families with unknown genetic basis	Scientific/ technological	Achieved
Identification of additional new predisposition genes for CRC in families with unknown genetic basis	Scientific/ technological	Foreseen within 2 years
Interactions between CRC susceptibility variants and environment	Scientific/ technological	Foreseen within 2 years
High risk CRC profile of inherited variants by statistical modelling	Scientific/ technological	Foreseen within 2 years
CRC profile used to identify individuals at a higher disease risk	Economic/Societal	Foreseen within 5-10 years
Genetic variants used to enhance CRC treatment efficacy	Economic/Societal	Foreseen within 5-10 years
CRC incidence reduction	Economic/Societal	Foreseen within 10+ years

I.E Dissemination and exploitation of Action results

Describe the Action's dissemination and exploitation approach as well as all activities undertaken to ensure dissemination and exploitation of Action results and the effectiveness of these activities.

Add description here

Item/ activity	Target audience	Result	Hyperlink
Website	Scientific community, health care workers, general population	Dissemination of activities and results	http://www.eucolongene.eu
Announcement at the European Journal of	Scientific community	Dissemination of Action to attract new members	http://www.nature.com/ejhg/index.html

5 Scientific/ technological, Economic, Societal
6 Achieved/ Foreseen within 2 years/ Foreseen 2-5 years/ Foreseen 5-10 years/ Foreseen 10+ years

Human Genetics			
Article	Scientific community	Identification of new CRC susceptibility variants	http://www.ncbi.nlm.nih.gov/pubmed/24743384
Article	Scientific community	Identification of new CRC susceptibility variants	http://www.ncbi.nlm.nih.gov/pubmed/24465592
Article	Scientific community	Identification of new CRC susceptibility variants	http://www.ncbi.nlm.nih.gov/pubmed/24218287
Article	Scientific community	Identification of new CRC susceptibility variants	http://www.ncbi.nlm.nih.gov/pubmed/24737748
Article	Scientific community	Identification of new CRC susceptibility variants	http://www.ncbi.nlm.nih.gov/pubmed/24978480
Article	Scientific community	Identification of new CRC susceptibility variants	http://www.ncbi.nlm.nih.gov/pubmed/26553438
Article	Scientific community	Identification of new CRC susceptibility variants	http://www.ncbi.nlm.nih.gov/pubmed/26621817
Article	Scientific community	Functional links for CRC susceptibility variants	http://www.ncbi.nlm.nih.gov/pubmed/25131200
Article	Scientific community	Genetic variants involved in CRC survival and treatment toxicity	http://www.ncbi.nlm.nih.gov/pubmed/23712746
Article	Scientific community	Genetic variants involved in CRC survival and treatment toxicity	http://www.ncbi.nlm.nih.gov/pubmed/25350395
Article	Scientific community	New predisposition genes for CRC in families with unknown genetic basis	http://www.ncbi.nlm.nih.gov/pubmed/23837913
Article	Scientific community	New predisposition genes for CRC in families with unknown genetic basis	http://www.ncbi.nlm.nih.gov/pubmed/24587672
Article	Scientific community	New predisposition genes for CRC in families with unknown genetic basis	http://www.ncbi.nlm.nih.gov/pubmed/25370038
Article	Scientific community	New predisposition genes for CRC in families with	http://www.ncbi.nlm.nih.gov/pubmed/25058500

		unknown genetic basis	
Article	Scientific community	New predisposition genes for CRC in families with unknown genetic basis	http://www.ncbi.nlm.nih.gov/pubmed/25860647
Workshop	Scientific community	Translational Genomics in Biomedicine-Barcelona	http://www.eucolongene.eu/translationalbiomed-workshop2014/
Workshop	Scientific community	NGS and cancer predisposition symposium-Leiden	http://www.eucolongene.eu
Workshop	Scientific community	Statistical Modeling of Cancer Genetic Predisposition-Vienna	http://www.eucolongene.eu
Workshop	Scientific community	Functional Genomics in Genetic Predisposition Studies-Istanbul	http://www.eucolongene.eu

I.F. Action success(es)

COST regularly communicates the successes of Actions. At this point in time what aspect(s) (outcomes and/ or impacts, rather than activities) of this Action is/ are the most suitable for communication?

Description of the success story	Dimension of the success ■ Breakthrough: scientific, technological or socioeconomic ■ Policy implementation (specify which policy) ■ Capacity building
Identification of new CRC susceptibility variants	Breakthrough: scientific, technological or socioeconomic
Identification of new predisposition genes for CRC	Breakthrough: scientific, technological or socioeconomic
Genome-wide association studies expertise including Inclusiveness Target Countries	Capacity building
Functional genomics expertise including Inclusiveness Target Countries	Capacity building
Next generation sequencing expertise including Inclusiveness Target Countries	Capacity building

II. Management Report

II.A. Overview of expenditure

Insert below in the yellow cells the summary of figures from the Yearly Financial Reports (YFRs) of completed Grant Periods and an IFR of any incomplete Grant Period – the Totals (non-yellow cells) will automatically sum.

[1] OERSA = Other Expenses Related to Scientific Expenditure (e.g. bank charges)
[2] FSAC = Amount received by Grant Holder for Financial Scientific and Administrative Coordination

II.B. Budget and Participation management

II.B.1 Budget spent in relation to individuals/ institutions outside participating COST countries

STSMs from or to institutions from countries other than Participating COST countries

The table below describes the added value STSMs to approved institutions in IPC or NNC or Specific Organisations and any STSMs from an approved institution in an NNC to a participating COST country.

Grantee		Host		Date	Topic and value added to the Action
Institution	Country	Institution	Country		
Add home institution and country		Add host institution and country		Date	Describe topic of the STSM and the added value to the Action
Add home institution and country		Add host institution and country		Date	Describe topic of the STSM and the added value to the Action
Add home institution and country		Add host institution and country		Date	Describe topic of the STSM and the added value to the Action

Invited Speakers

The table below highlights the added value of Invited Speakers from COST countries that have not accepted the MoU and/ or non-participating NNC, IPC or Specific Organisations whose participation at a meeting or Training School was reimbursed by the Action.

Participant name	Institution	Country	Event date	Topic and added value to the Action
Luis Carvajal-Carmona	University of California Davis	US	12/03/2014	Cancer genetics in Latin America. Luis Carvajal-Carmona presented his research on cancer genetics at the University of California, Davis.

				Luis is a former member of Ian Tomlinson's group and he is now an assistant professor at this university. He is continuing his previous research in CRC genetics and he was on the process to join officially our Action (he did later on in 03/05/2014).

Dissemination meetings

The table below highlights the added value of Dissemination Meetings financed from Action funds.

Participant name	Role	Country	Date	Location	Topic and added value to the Action
Add	Add	Add	Add	Add	Describe the speaker's topic and the added value to the Action

II.C. Participants

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II.D. Specific issues

This section is confidential to the Management Committee, and the COST Association (Administration, Scientific Committee and Committee of Senior Officials); and is not included in the version of the report that is published on the COST website.

The Action encountered the following particular difficulties in the implementation of the Action (e.g. imbalances of participation across the Working Groups, inactive country representatives).

This Action was based on a previously existing consortium (COGENT, COlorectal cancer GENeTics, http://www.ncbi.nlm.nih.gov/pubmed/19920828, http://www.ncbi.nlm.nih.gov/pubmed/22294761). This fact can be considered positive in general but it may have brought some differences between former members and new participants joining after 30/04/2013. There has been a good general participation so far of all members in activities organized by this Action including workshops, meetings, training school, and STSMs. However, it can be stated in general that former participants have been more active regarding their participation in research projects developed jointly. Some reasons are listed below:

- Existing differences regarding CRC research stage (more advanced for old, less advanced for new). In some cases, new groups started collecting samples when joining this Action just to mention a clear example.
- Lack of funding to develop CRC projects to follow this Action's objectives (ex. no funding to perform GWAS or NGS).
- Action's objectives do not match well with their own research
- Own research objectives shift into other topics different from CRC

Finally, it should be mentioned that some participants never attended any activity (Sanja Kapitanovic HR, Pierre Ellul MT, Rolf Sijmons NL, Cristian Lupascu RO, Anca Trifan RO, Angel Carracedo ES, Tao Liu SE).

Describe the issue(s) here or write "no particular difficulties encountered".

The MC did not accept the pending intentions to accept the MoU shown in Section I.A for the following reason.

There are no pending intentions to join this Action.

Complete list of participants

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Annex 1

Definitions:

COST Action Challenge (main aim)	"The research question addressed by the COST Action targeting scientific, technological, and / or socioeconomic problems"
COST Action Innovation	"The creation and / or development of new or improved concepts, products, processes, services, and / or technologies that are made available to markets, governments and society"
COST Action objectives	"COST Action objectives are the results that an Action needs to achieve in order to respond to meet its challenge. These are SMART (Specific, Measurable, Achievable, Relevant, Timely) and twofold: research coordination objectives and capacity building objectives."
COST Action research coordination objectives	"Achieving these objectives turns COST Actions from initially scattered teams into one transnational team and leverages the existing funded research. These objectives entail the distribution of tasks, sharing of knowledge and know-how, and the creation of synergies among Action participants to achieve specific outputs."
COST Action capacity building objectives	"Achieving these objectives entail building critical mass to drive scientific progress, thereby strengthening the European Research Area. They can be achieved by the delivery of specific outputs and / or through network features or types and levels of participation."
COST Action networking activities	"any activities organised by the COST Action (whether or not directly funded by COST) in order to achieve research coordination and capacity building objectives."
COST Action networking tools	"instruments through which eligible activities can be funded"
COST Action outputs	"direct results from the COST Action activities. These can be codified knowledge, tacit knowledge, technology, and societal applications."
COST Action impact	"the short- to long-term scientific, technological, and / or socioeconomic changes produced by a COST Action, directly or indirectly, intended or unintended."
COST Action deliverable	"a distinct, expected and tangible output of the Action, meaningful in terms of the Action's overall objectives such as a report, a document, a technical diagram, a software etc. Action deliverables are used to measure its progress and success."
COST Action milestones	"Control points in the Action that help to chart progress. They are also needed at intermediary points so that, if problems have arisen, corrective measures can be taken. A milestone may be a critical decision point in the Action where, for example, the MC must decide which of several technologies to adopt for further development (e.g. core group and MC meetings, mid-term reviews)"
Inclusiveness Target Country (ITC):	Current COST Member Countries targeted by the COST inclusiveness Policy ("Inclusiveness Target Countries" (ITC)): EU 13 (Bulgaria, Cyprus, Czech Republic, Estonia, Croatia, Hungary, Lithuania, Latvia, Malta, Poland, Romania, Slovenia, Slovakia), EU candidate countries (the former Yugoslav Republic of Macedonia, Montenegro, Republic of Serbia, Turkey) and potential EU candidate countries (Bosnia and Herzegovina). In addition, to comply with the EC criteria for 'Spreading Excellence and Widening Participation', Portugal and Luxemburg are included.