Gemelli

Webinar Italynch 21 ottobre 2020

Mainstreaming: pro e contro rispetto al modello tradizionale

Il punto di vista *di un* genetista

Medicina Genomica
Dipartimento di Scienze della Vita e Sanità Pubblica
Università Cattolica del Sacro Cuore

UOC Genetica Medica,
Fondazione Policlinico Universitario "A. Gemelli"
Roma

Recommendations from the EGAPP Working Group: genetic testing strategies in newly diagnosed individuals with colorectal cancer aimed at reducing morbidity and mortality from Lynch syndrome in relatives

*Evaluation of Genomic Applications in Practice and Prevention (EGAPP) Working Group**

VOLUME 31 · NUMBER 20 · JULY 10 2013
JOURNAL OF CLINICAL ONCOLOGY ORIGINAL REPORT

Population-Based Molecular Screening for Lynch Syndrome: Implications for Personalized Medicine

Robyn L. Ward, Sian Hicks, and Nicholas J. Hawkins

VOLUME 31 · NUMBER 20 · JULY 10 2013
JOURNAL OF CLINICAL ONCOLOGY EDITORIAL

Population-Based Universal Screening for Lynch Syndrome: Ready, Set… How?

lentification of Lynch Syndrome
mong Patients With Colorectal Cancer

MA, October 17, 2012—Vol 308, No. 15

Annals of Oncology 25: 2001–2007, 2014
doi:10.1093/annonc/mdu361
Published online 31 July 2014

rfold increased detection of Lynch syndrome
aising age limit for tumour genetic testing from
o 70 years is cost-effective

ie[1], A. R. Mensenkamp[1], E. M. M. Adang[2], M. J. L. Ligtenberg[1,3] & N. Hoogerbrugge[1*]

RESEARCH ARTICLE

Cost-effectiveness analysis of genetic diagnostic strategies for Lynch syndrome in Italy

Roberta Pastorino[1]*, Michele Basile[2], Alessia Tognetto[3], Marco Di Marco[4], Adriano Grossi[3], Emanuela Lucci-Cordisco[5,6], Franco Scaldaferri[7], Andrea De Censi[8], Antonio Federici[9], Paolo Villari[4], Maurizio Genuardi[5,6], Walter Ricciardi[1,3], Stefania Boccia[1,3]

PLOS ONE | https://doi.org/10.1371/journal.pone.0235038 July 1, 2020

Universal tumor testing for Lynch syndrome

- Dimostrate:
  - ➢Validità analitica
  - ➢Validità clinica
  - ➢Utilità clinica

- Cost-effective

Recommendations from the EGAPP Working Group: genetic testing strategies in newly diagnosed individuals with colorectal cancer aimed at reducing morbidity and mortality from Lynch syndrome in relatives

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RESEARCH ARTICLE

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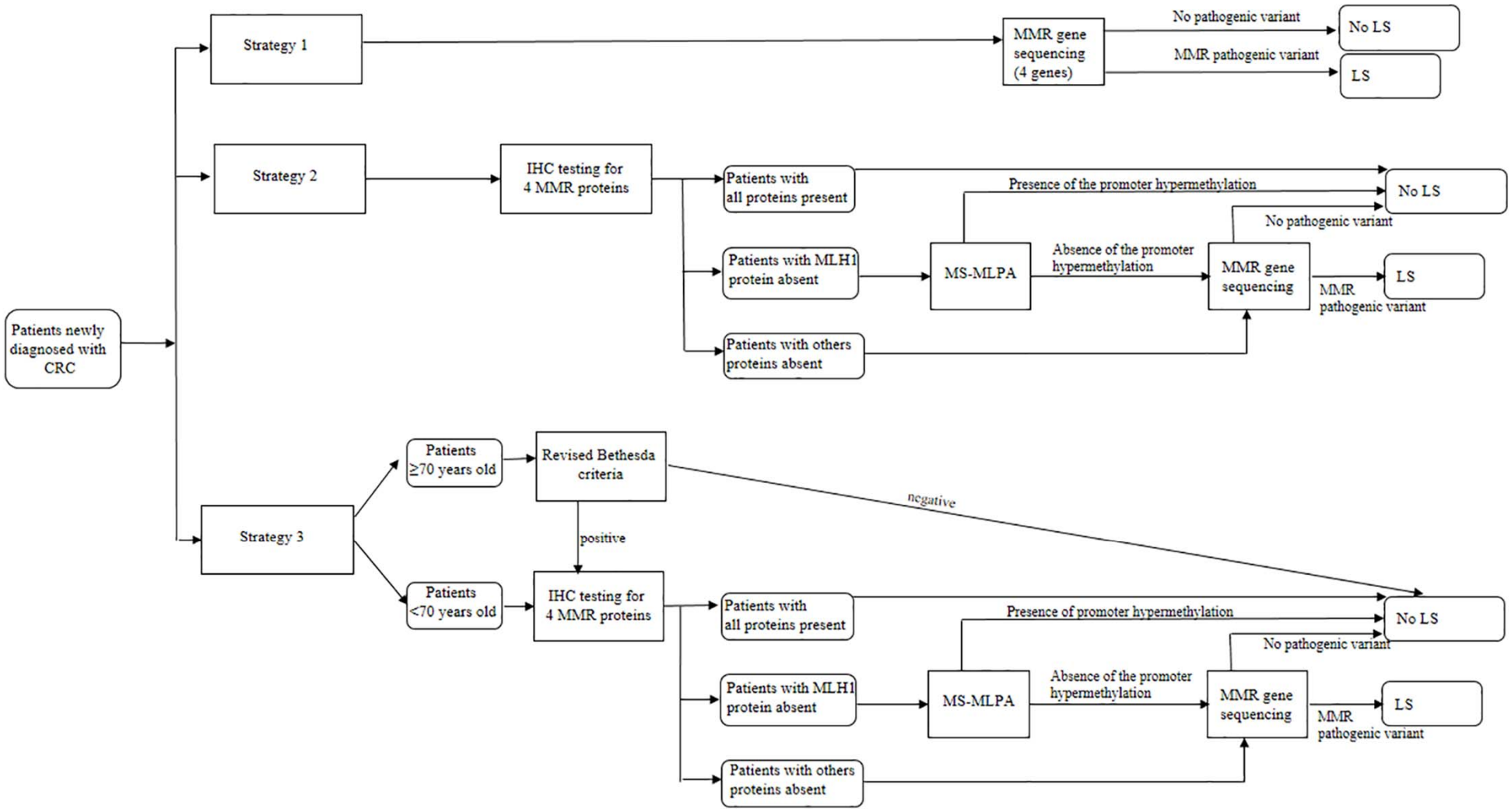
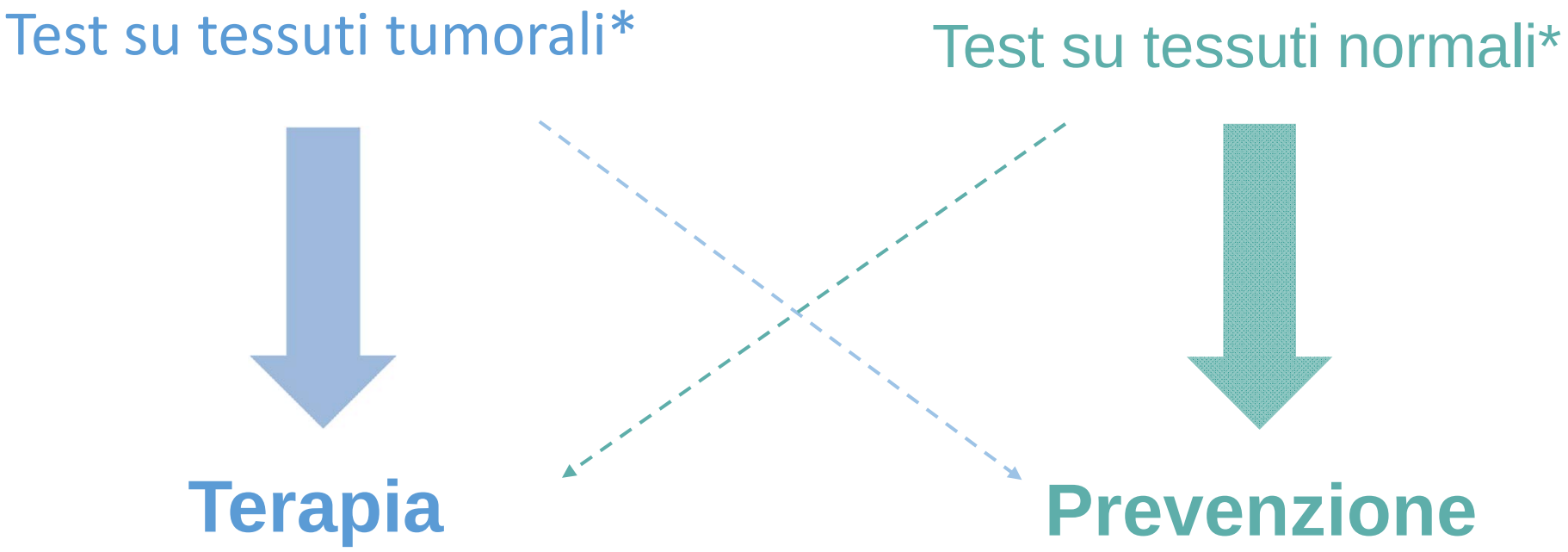
PLOS ONE | https://doi.org/10.1371/journal.pone.0235038 July 1, 2020


Patients newly diagnosed with CRC
Strategy 1
MMR gene sequencing (4 genes)
No pathogenic variant
No LS
MMR pathogenic variant
LS
Strategy 2
IHC testing for 4 MMR proteins
Patients with all proteins present
Presence of the promoter hypermethylation
No LS
No pathogenic variant
Patients with MLH1 protein absent
MS-MLPA
Absence of the promoter hypermethylation
MMR gene sequencing
LS
MMR pathogenic variant
Patients with others proteins absent
Strategy 3
Patients ≥70 years old
Revised Bethesda criteria
negative
positive
Patients <70 years old
IHC testing for 4 MMR proteins
Patients with all proteins present
Presence of promoter hypermethylation
No LS
No pathogenic variant
Patients with MLH1 protein absent
MS-MLPA
Absence of the promoter hypermethylation
MMR gene sequencing
LS
MMR pathogenic variant
Patients with others proteins absent

Finalità dei test genetici in oncologia



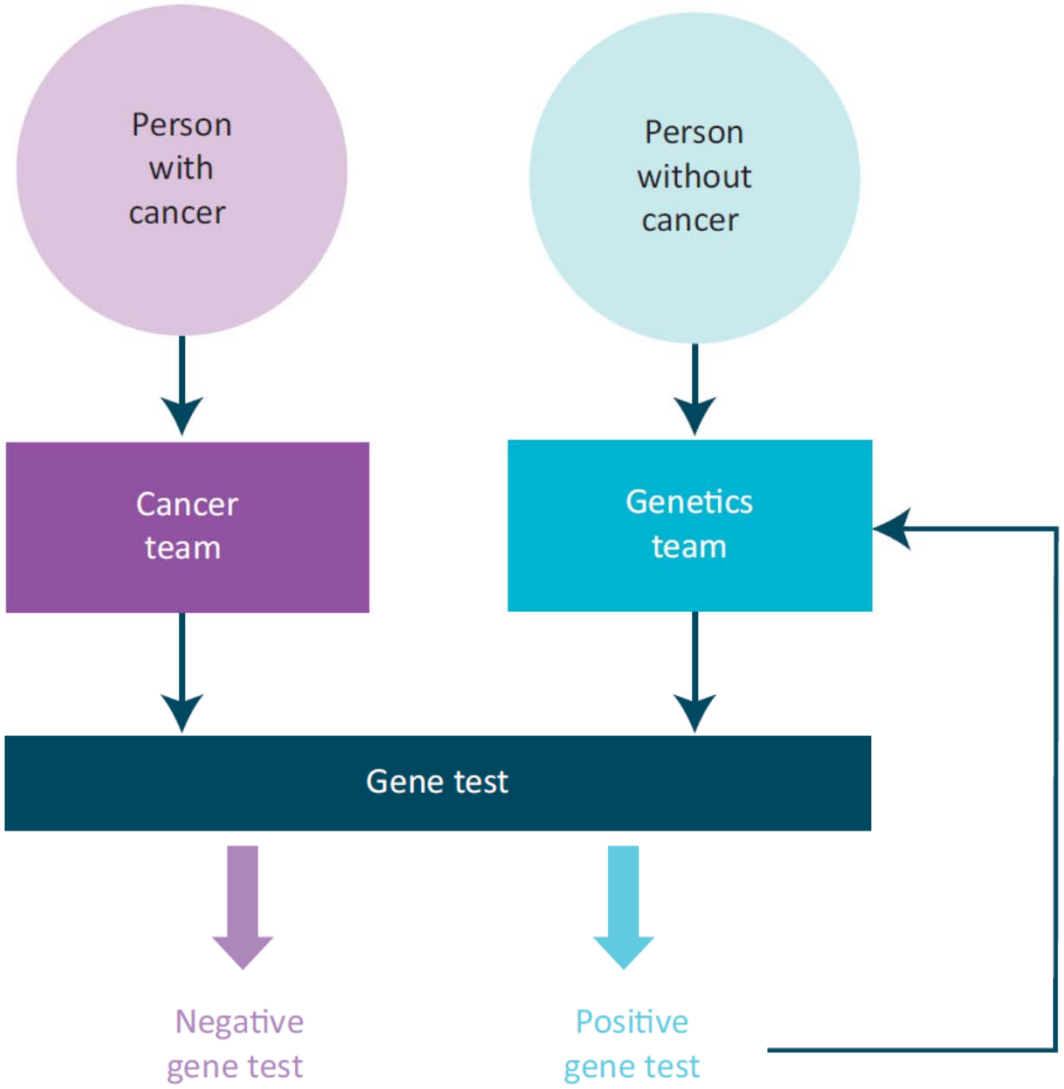
*Test *somatici*
(campioni inclusi in paraffina)

*Test *costituzionali* o *germinali*
(sangue, altro)

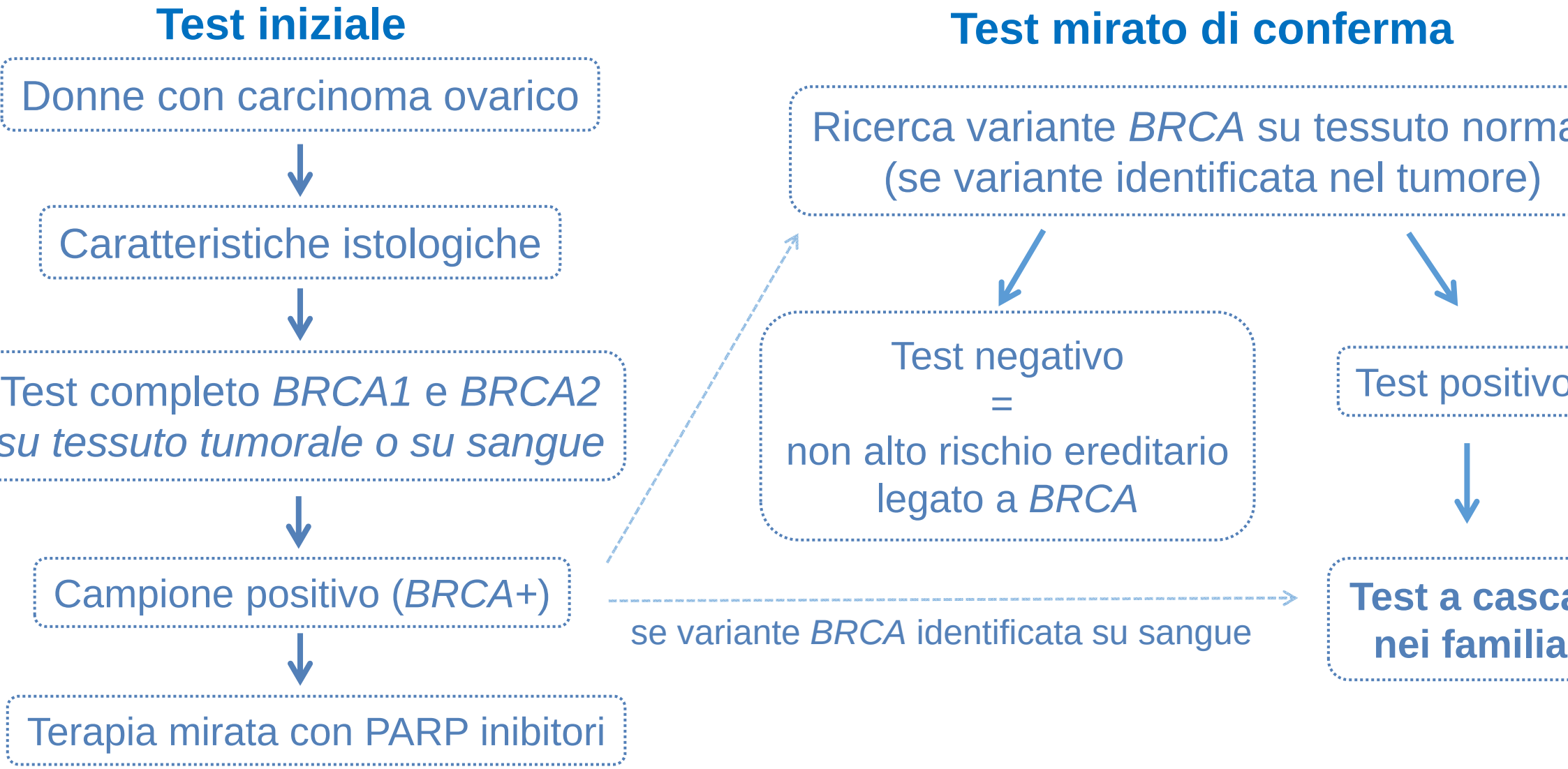
Clinical Medicine 2014 Vol 14, No 4: 436–9

Mainstreaming genetic testing of cancer predisposition genes

Author: Nazneen Rahman[A]



Identificazione donne BRCA+: percorso terapeutico



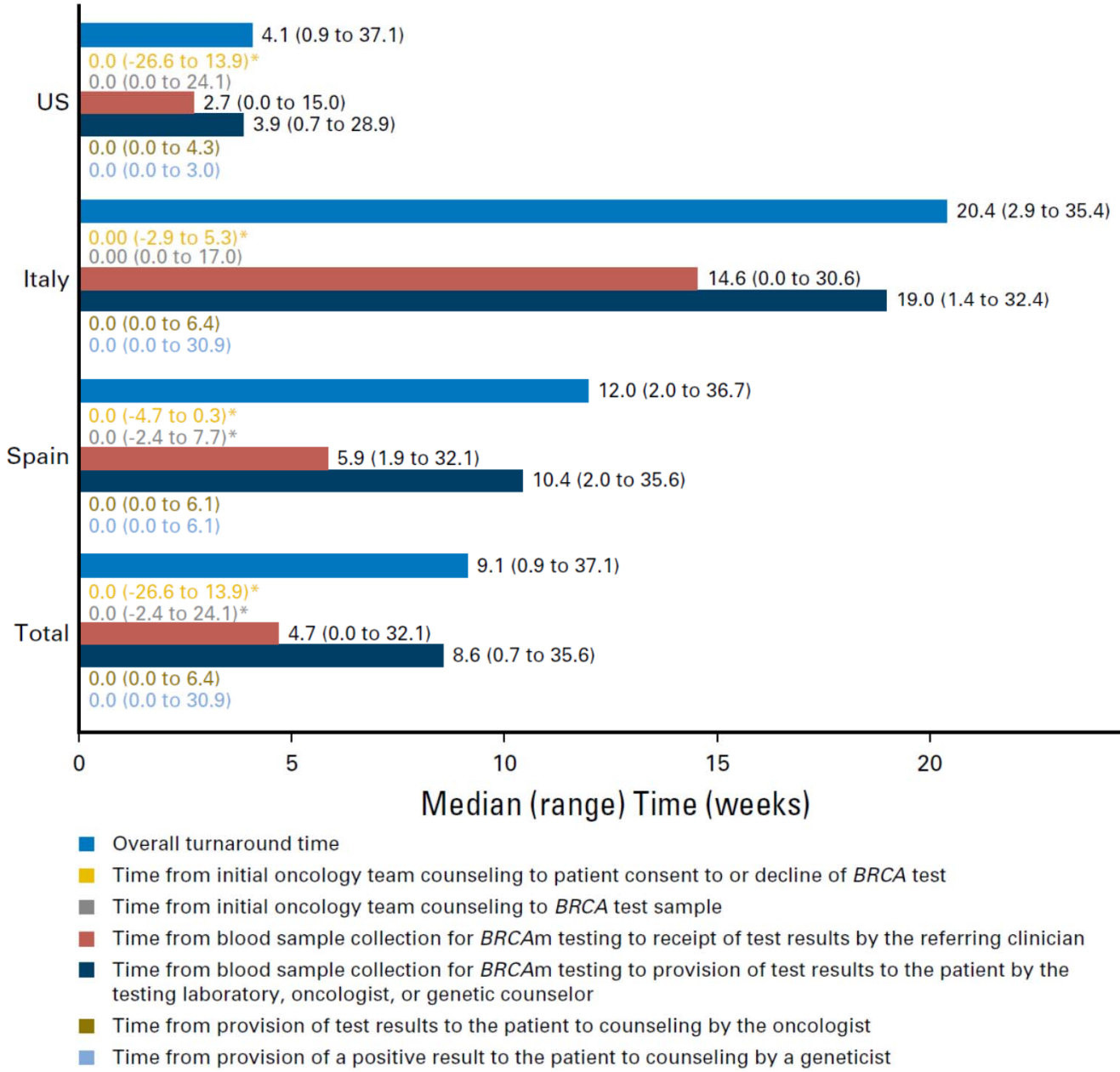
· NUMBER 13 · MAY 1, 2018

L OF CLINICAL ONCOLOGY

ORIGINAL REPORT

Evaluation of a Streamlined Oncologist-Led *BRCA* Mutation Testing and Counseling Model for Patients With Ovarian Cancer

Nicoletta Colombo, Gloria Huang, Giovanni Scambia, Eva Chalas, Sandro Pignata, James Fiorica, Linda Van Le, Sharad Ghamande, Santiago González-Santiago, Isabel Bover, Begoña Graña Suárez, Andrew Green, Philippe Huot-Marchand, Yann Bourhis, Sudeep Karve, and Christopher Blakeley



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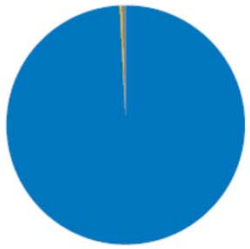
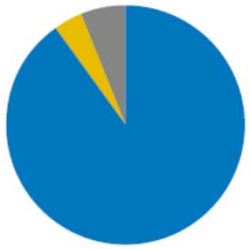
Did your oncologist or nurse discuss: Yes No I don't remember

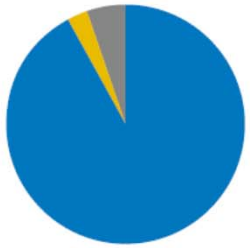
The purpose of the *BRCA* test
Q1



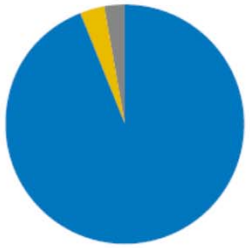
How the *BRCA* test might affect your treatment
Q2



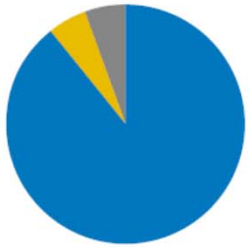
The *BRCA* gene and its function
Q3



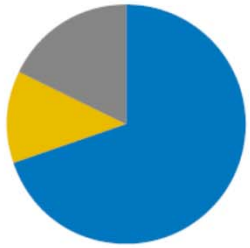
The implications of a positive test result
Q4



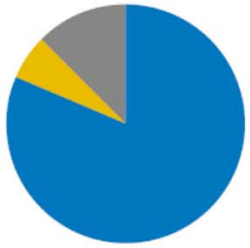
The implications of a negative test result
Q5



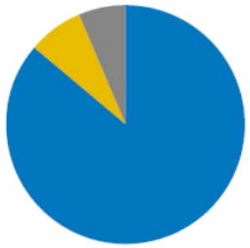
The implications of a VUS
Q6



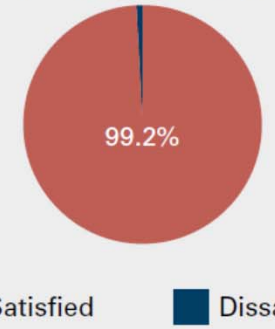
The accuracy of the *BRCA* test
Q7



Your feelings about the *BRCA* test
Q10



In summary:
How would you rate your satisfaction with the counseling?



Cancer genetics, precision prevention and a call to action

Clare Turnbull[1,2,3,4*], Amit Sud[1] and Richard S. Houlston[1]

NATURE GENETICS | VOL 50 | SEPTEMBER 2018 | 1212–1218 |

fully deliver a precision prevention program, long-term, large-scale mutation studies that capture longitudinal clinical data
d serial biosamples are required.

VANTAGGI DEL MAINSTREAMING

- Finalità terapeutica
- Tempistica
- Accesso universale
- Maggior sensibilità
- Prevenzione su larga scala

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PROBLEMI ASSOCIATI AL MAINSTREAMING

- Consenso per test MMRd?
- Contenuto informativo
- Comprensione
- Invio al genetista
- Definizione rischi
- Quali strategie preventive

Nonostante siano state prodotte evidenze di soddisfazione degli utenti per modalità di (teleconsulenza) e mainstreaming, i dati di confronto sono molto limitati

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ALLEANZA CONTRO IL CANCRO

GerSom

Studio di fattibilità per la diagnosi genomica congiunta di rischio genetico e di sensibilità ai nuovi farmaci nelle neoplasie del seno, ovaio e colon

Abstract: Il programma di ricerca si pone quale obiettivo generale lo studio della fattibilità di un percorso diagnostico congiunto, al momento della diagnosi di tumore, per la identificazione contemporanea delle mutazioni somatiche (a scopi prognostici e di definizione della risposta alla terapia) e delle varianti ereditarie (per la identificazione del rischio genetico di tumore) in pazienti con tumori dell'ovaio, alcuni sottotipi di tumore della mammella (triplo-negativi) o tumori del colon/retto. La fattibilità di questo progetto comporta due elementi principali: i) l'allestimento di un pannello di geni per analisi simultanea delle varianti ereditarie e delle mutazioni somatiche, a costi contenuti (<400€) ed eseguibile in tempi brevi (<1 settimana); ii) l'allestimento di un database di informazioni scientifiche che consenta la gestione clinica corretta delle informazioni derivanti dall'analisi genetica (sia per la scelta dei trattamenti che per la mappatura del rischio genetico).