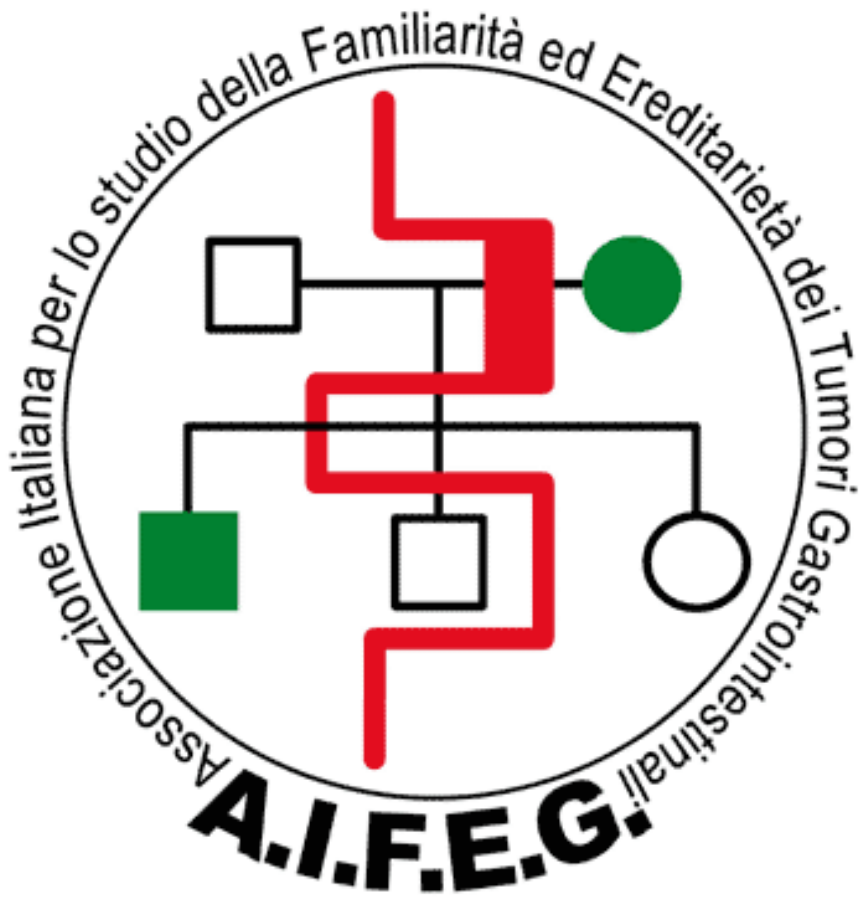

Associazione Italiana per lo Studio della Familiarità ed Ereditarietà dei Tumori Gastrointestinali

G.N. Ranzani: ***guglielmina.ranzani@unipv.it***

A.I.F.E.G. - history of the Association

Cristina Oliani

- the A.I.F.E.G. was officially founded in Verona in July 2002
- but it can be traced back to “the Italian study group on hereditary colorectal cancers” established in 1993
  - the group included physicians, as well as clinical and basic researchers from different institutions all over the country
  - since 1993 the group organized an annual national meeting in order to encourage the exchange of information, clinical experiences and scientific thoughts

The founding fathers of the Italian study group

Maurizio Ponz de Leon
(University of Modena)

Lucio Bertario
(Cancer Institute-INT, Milano)

A.I.F.E.G. - Aims

- To promote:
  - research activity on genetic factors predisposing to gastrointestinal tumors
  - research activity on tumor prevention and treatment
  - collaborative projects on basic, pre-clinical, and clinical research
  - exchanges and collaborations for closing the gaps from knowledge to practice and putting knowledge into action
  - dissemination of the acquired knowledge at different levels

A.I.F.E.G.

- was the first Italian association aimed at investigating hereditary gastrointestinal tumors
- was conceived as a multidisciplinary resource for studying hereditary gastrointestinal tumors thoroughly, optimizing diagnosis and treatment
- was founded in the context of the Italian health system (not always ready to accept a multidisciplinary approach in health research and services)
- in spite of some difficulties and challenges, it has become a reference point for medical geneticists and physicians of different disciplines to understand better how to manage patients and families at risk

AIFEG

Associazione Italiana per lo Studio della Familiarità ed Ereditarietà dei Tumori Gastrointestinali

PROSSIMI EVENTI

CENTRI AIFEG

COME ASSOCIARSI

CONTATTI

2017

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DOI 10.1186/s13148-015-0165-2

Collaborative studies

Clinical Epigenetics

RESEARCH Open Access

Aberrant DNA methylation profiles of inherited and sporadic colorectal cancer

Nora Sahnane[1], Francesca Magnoli[1], Barbara Bernasconi[1], Maria Grazia Tibiletti[2], Chiara Romualdi[3], Monica Pedroni[4], Maurizio Ponz de Leon[4], Giulia Magnani[4], Luca Reggiani-Bonetti[5], Lucio Bertario[6], Stefano Signoroni[6], Carlo Capella[1], Fausto Sessa[1], Daniela Furlan[1]* and AIFEG

PATHOLOGICA 2015;107:104-109

Collaborative studies

REVIEW

Immunohistochemical evaluation of mismatch repair proteins in colorectal carcinoma: the AIFEG/GIPAD proposal

A. REMO[1*], M. FASSAN[2*], G. LANZA[1,2] ON BEHALF OF AIFEG AND GIPAD

[1] Italian Association studying Familial and Hereditary Gastrointestinal Tumors (AIFEG): Chair: GB. Rossi. Directive members: D.Barana, B. Bonanni, M. Pedroni, A. Remo, E.D. Urso, M. Vitellaro; [2] Italian Group of Gastrointestinal Pathologists (GIPAD): Chairs: M. Guido & L. Saragoni. Scientific committee: F.P. D'Armiento, M. Fassan, L. Mastracci

[*] These authors contributed equally to this work.

Collaborative studies

Digestive and Liver Disease 45 (2013) 606–611

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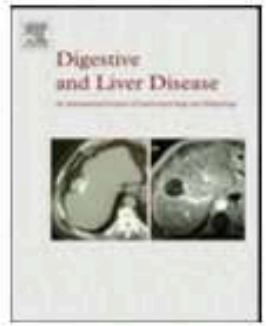

Oncology

Cancer risk associated with *STK11/LKB1* germline mutations in Peutz–Jeghers syndrome patients: Results of an Italian multicenter study

Nicoletta Resta[a,*], Daniela Pierannunzio[b], Gennaro Mariano Lenato[c], Alessandro Stella[a], Riccardo Capocaccia[b], Rosanna Bagnulo[a], Patrizia Lastella[c], Francesco Claudio Susca[a], Cristina Bozzao[a], Daria Carmela Loconte[a], Carlo Sabbà[c,d], Emanuele Urso[e], Paola Sala[f], Mara Fornasarig[g], Paola Grammatico[h], Ada Piepoli[i], Cristina Host[j], Daniela Turchetti[k], Alessandra Viel[l], Luigi Memo[m], Laura Giunti[n], Vittoria Stigliano[o], Liliana Varesco[p], Lucio Bertario[f], Maurizio Genuardi[q,r], Emanuela Lucci Cordisco[s], Maria Grazia Tibiletti[t], Carmela Di Gregorio[u], Angelo Andriulli[i], Maurizio Ponz de Leon[v], AIFEG

J Med Genet. 2013 Dec;50(12):785-93. doi: 10.1136/jmedgenet-2013-101803. Epub 2013 Aug 16.

Criteria and prediction models for mismatch repair gene mutations: a review.

Win AK[1], Macinnis RJ, Dowty JG, Jenkins MA.

Author information

Abstract

One of the strongest predictors of colorectal cancer risk is carrying a germline mutation in a DNA mismatch repair (MMR) gene. Once identified, mutation carriers can be recommended for intensive screening that will substantially reduce their high colorectal cancer risk. Conversely, the relatives of carriers identified as non-carriers can be relieved of the burden of intensive screening. Criteria and prediction models that identify likely mutation carriers are needed for cost-effective, targeted, germline testing for MMR gene mutation. We reviewed 12 criteria/guidelines and 8 prediction models (Leiden, Amsterdam-plus, Amsterdam-alternative, MMRpro, PREMM1,2,6, MMRpredict, Associazione Italiana per lo studio della Familiarità ed Ereditarietà dei tumori Gastrointestinali (AIFEG) and the Myria
to identify individuals with colorectal ca
MMRpredict and Myriad tables can pre
We conducted a meta-analysis of the d
estimate for the area under curve was
(95% CI 0.81 to 0.88) for PREMM, and
we cannot state that one model has a h
been shown to be sensitive and specifi
provide prediction of PMS2 mutations,
family history, and be applicable to all p

Collaborative studies

Clin Genet. 2006 Mar;69(3):254-62.

A genetic model for determining MSH2 and MLH1 carrier probabilities based on family history and tumor microsatellite instability.

Marroni F[1], Pastrello C, Benatti P, Torrini M, Barana D, Cordisco EL, Viel A, Mareni C, Oliani C, Genuardi M, Bailey-Wilson JE, Ponz de Leon M, Presciuttini S.

Author information

Abstract

Mutation-predicting models can be useful when deciding on the genetic testing of individuals at risk and in determining the cost effectiveness of screening strategies at the population level. The aim of this study was to evaluate the performance of a newly developed genetic model that incorporates tumor microsatellite instability (MSI) information, called the AIFEG model, and in predicting the presence of mutations in MSH2 and MLH1 in probands with suspected hereditary non-polyposis colorectal cancer. The AIFEG model is based on published estimates of mutation frequencies and cancer penetrances in carriers and non-carriers and employs the program MLINK of the FASTLINK package to calculate the proband's carrier probability. Model performance is evaluated in a series of 219 families screened for mutations in both MSH2 and MLH1, in which 68 disease-causing mutations were identified. Predictions are first obtained using family history only and then converted into posterior probabilities using information on MSI. This improves predictions substantially. Using a probability threshold of 10% for mutation analysis, the AIFEG model applied to our series has 100% sensitivity and 71% specificity.

Convivio sull'Ereditarietà e XIV Congresso AIFEG

Associazione Italiana per lo Studio delle Familiarità ed Ereditarietà nei Tumori Gastrointestinali

Verona

10-11-12 Novembre 2016

CENTRO MEDICO MARANI

Dead Line abstract: 20 ottobre 2016

Inviare gli abstract al seguente indirizzo: ANDREA.REMO@AULSSLEGNAGO.IT

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Very recently

- a PATIENTS ASSOCIATION linked to A.I.F.E.G. was set up by a Lynch patient who was motivated by personal events
- this link helped in obtaining prescription charge exemption for Lynch genetic test and cancer prevention (including specialized cancer screening, intensified surveillance, and/or prophylactic surgeries)
- dissemination and educational activities are progressively enhanced through linkages with other Scientific Associations such as SIGU, SIC……

IEO education
Istituto Europeo di Oncologia

RIUNIONI DI GENETICA CLINICA

30/06/2017 ORE 13.00 – 14.00
IEO – AULA B

“SINDROME DI LYNCH: NOVITA’ GENETICHE E ATTUALE APPROCCIO CLINICO”

13.00 **B. BONANNI**	*BENVENUTO E INTRODUZIONE*
13.05 **L. BERTARIO**	*QUANDO SOSPETTARLA CLINICAMENTE?*
13.20 **M.G. TIBILETTI**	*NUOVE MODALITA’ DI APPROCCIO DIAGNOSTICO*
13.40	*CASI CLINICI E DISCUSSIONE GENERALE*

In collaborazione con

myriad 25 YEARS WHEN DECISIONS MATTER

Patrocinio richiesto a

SIGU web
Società Italiana di Genetica Umana

Associazione Italiana per lo studio della Familiarità ed Ereditarietà dei Tumori Gastroenterologici A.I.F.E.G.

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Educational

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Alessandra Viel	*Cancer Institute (CRO)-Aviano*

There is still a long way to go……

Thank you for your attention and

please bear AIFEG in mind

for future collaborations