Programme – InSiGHT 2015 – Sao Paulo – Brazil

June 18th 2015 - Thursday

7:00 – Registration Opens

8:00 – Official Opening

Benedito Mauro Rossi - Chairman
David Uip – Secretary of Health – São Paulo State - Brazil

8:05 - Session 1
Chairpersons
Hans Vasen – Finlay Macrae

>Henry Lynch Lecture (20 minutes)
  Introduction by Patrick Lynch
  What epidemiology tells us about Lynch Syndrome
  Mark Jenkins

>Oral presentations (60 minutes)
(presentation: 7 minutes + questions: 3 minutes)

103 - IMPACT OF COLONOSCOPY ON RISK OF COLORECTAL CANCER FOR MEMBERS OF LYNCH SYNDROME FAMILIES
DRISS AIT OUAKRIM

6 - VACCINATION WITH MONOCYTE-DERIVED DENDRITIC CELLS IN LYNCH SYNDROME PATIENTS: VIGOROUS T CELL RESPONSES TO NEOANTIGEN FRAMESHIFT-DERIVED PEPTIDES
NICOLINE HOOGERBRUGGE

130 - COPY NUMBER VARIATION ANALYSIS IN 85 SUSPECTED LYNCH SYNDROME FAMILIES REVEALS NOVEL POTENTIAL CAUSATIVE CANDIDATE GENES
KATRIN KAYSER
13 - PROSPECTIVE CANCER RISKS AND SURVIVAL IN HEALTHY MMR MUTATION CARRIERS SUBJECT TO SURVEILLANCE COLONOSCOPY
PÅL MØLLER

85 - BI-ALLELIC SOMATIC MUTATIONS AS A CAUSE OF TUMOUR MISMATCH REPAIR-DEFICIENCY IN COLORECTAL CANCER: IMPLICATIONS FOR IDENTIFYING MISMATCH REPAIR GENE MUTATION CARRIERS WITHIN POPULATION-BASED COLORECTAL CANCER
DANIEL BUCHANAN

49 - CANCER RISKS IN FAMILY MEMBERS OF CMMR-D PATIENTS
MAARTJE NIELSEN

>Review (20 minutes)
Hereditary Breast and Colorectal Cancer
Annika Lindblom

10:00 - Break (including electronic posters)

10:30 - Session 2
Chairpersons
Allan Spigelman – Mark Jenkins

>Review (20 minutes)
Hereditary Diffuse Gastric Cancer
Susan Parry

>Oral presentations (60 minutes)
(presentation: 7 minutes + questions: 3 minutes)

55 - EVIDENCE OF INFLUENCE OF ASPIRIN ON MUCOSAL IMMUNE STATUS AND ON THE CARCINOGENIC EFFECTS OF OBESITY SUPPORT THE NEED FOR THE DOSE NON-INFERIORITY STUDY, CAPP3
JOHN BURN

198 - BETTER EDUCATION IS NEEDED FOR BOTH HNPCC FAMILY MEMBERS AND THEIR PROVIDERS
DENNIS JAMES AHNEN

16 - EXTRACOLONIC CANCER IN Lynch SYNDROME
CHRISTINA THERKILDSEN

190 - YEARLY GASTROSCOPY IN MLH1 AND MSH2 MUTATION CARRIERS - AN ENDOSCOPY TOO FAR?
SUSAN PARRY

76 - IDENTIFYING LYNCH SYNDROME USING UNIVERSAL COLORECTAL CANCER SCREENING: IMPLICATIONS OF PATIENT AGE
MATTHEW F KALADY

14 - TIME BETWEEN COLONOSCOPY, COLORECTAL CANCER INCIDENCE AND DEATH IN MLH1 MUTATION CARRIERS
PÅL MØLLER

> Aldred Scott Warthin Lecture (20 minutes)
  Introduction by Matthew Kalady

  The MMR system: from bench to bedside
  Gabriel Capellá

12:30 – Lunch (including electronic posters)

12:45 – 13:45
InSiGHT Council Meeting & Lunch (by invitation only)

14:00 - Session 3
Chairpersons
Annika Lindblom – James Church

> Review (20 minutes)
  GI cancers in Li-Fraumeni Syndrome
  Maria Isabel W. Achatz

> Oral presentations (60 minutes)
  (presentation: 7 minutes + questions: 3 minutes)

201 - RESULTS OF HIGH/MODERATE CANCER GENE PANEL TESTS IN AN ETHNICALLY DIVERSE PATIENT POPULATION
MONICA M. ALVARADO
138 - EXOME SEQUENCING OF AN AMSTERDAM-POSITIVE FAMILY IDENTIFIES A NOVEL CAUSAL GENE FOR HEREDITARY NON-POLYPOSIS COLORECTAL CANCER
LAURA VALLE

95 - SPECTRUM OF CANCER PHENOTYPES IN ASIAN LYNCH SYNDROME FAMILIES.
CHUN GAN

106 - UPDATING THE INSIGHT DATABASE TO MEET THE CHALLENGES OF THE GENOME SEQUENCING ERA
JOHN-PAUL PLAZZER

45 - METACHRONOUS COLORECTAL CANCER IN A GENERAL NATIONAL COHORT FROM 1943-2012 AND ITS RELEVANCE AS INDICATOR OF HEREDITARY COLORECTAL CANCER
LARS JOACHIM LINDBERG

98 - SHORT-TERM RISK OF COLORECTAL CANCER FOR LYNCH SYNDROME: A META-ANALYSIS
MARK JENKINS

>Meera Khan Lecture (20 minutes)
Introduction by Thomas Weber

Towards universal screening for Lynch Syndrome
Ian Frayling

16:00 - Break (including electronic posters)
16:30 - Session 4
Chairpersons
Patrick Lynch – Gabriela Moeslein

>Review (20 minutes)
Chemotherapy for Hereditary GI Cancer
Paulo Hoff

>Oral presentations (25 minutes)
(presentation: 2 minutes + questions: 1 minute)

81 - MLH1 MUTATION TYPE AND FREQUENCY IN COLORECTAL CARCINOMAS DEMONSTRATING SOLITARY LOSS OF PMS2 PROTEIN EXPRESSION
DANIEL BUCHANAN
137 - SPECIFIC BACTERIAL SEQUENCES DETERMINATION IN FECES IDENTIFIES HIGHER COLORECTAL NEOPLASIA RISK SUBGROUP AMONG LYNCH SYNDROME CARRIERS
GABRIEL CAPELLÁ

97 - VALIDATION OF LYNCH SYNDROME PREDICTION MODELS IN ASIAN POPULATIONS
CHUN GAN

129 - COMMON GENETIC VARIANTS WITHIN THE TERT GENE AND RISK OF COLORECTAL CANCER FOR DNA MISMATCH REPAIR GENE MUTATION CARRIERS
DANIEL BUCHANAN

160 - UPTAKE OF GENETIC TESTING AMONG RELATIVES OF LYNCH SYNDROME CARRIERS IN A UNITED STATES CANCER GENETICS REGISTRY
ELENA MARTINEZ STOFFEL

172 - GERMLINE MLH1 MUTATIONS IN INDIVIDUALS WITH PMS2 DEFICIENT TUMOURS
KARA SEMOTIUK

57 - THE FORGOTTEN GI CANCERS IN FAP
SARAH-JANE YVONNE WALTON

31 - ACTIVATED SYSTEMIC DENDRITIC CELL PHENOTYPE IN FAMILIAL ADENOMATOUS POLYPOSIS (FAP) – DOES APC MUTATION AFFECT THE ANTIGEN PRESENTING CELLS OF THE INNATE IMMUNE SYSTEM?
GUI HAN LEE

>Discussion of selected cases – interactive (40 minutes)
  Francisco Lopez – Carlos Vaccaro
  Moderated by Patricia Prolla

18:30 - Reception
June 19th 2015 - Friday

8:00 - Session 5
Chairpersons
Maurizio Genuardi – John Paul Plazzer

Sir Ian Todd Lecture (20 minutes)
Introduction by Susan Clark

New colorectal cancer predisposition genes identified by NGS
Nicoline Hoogerbrugge

Oral presentations (60 minutes)
(presentation: 7 minutes + questions: 3 minutes)

12 - RISK OF EXTRACOLONIC CANCERS FOR CARRIERS OF BIALLELIC AND MONOALLELIC MUTATIONS IN MUTYH
AUNG KO WIN

7 - FREQUENCY AND PHENOTYPIC SPECTRUM OF GERMLINE MUTATIONS IN POLE AND SEVEN OTHER POLYMERASE GENES IN 266 PATIENTS WITH COLORECTAL ADENOMAS AND CARCINOMAS
ISABEL SPIER

116 - POLYMERASE PROOFREADING-ASSOCIATED SYNDROME: POLE AND POLD1 MUTATIONS IN HEREDITARY COLORECTAL CANCER AND POLYPOSIS
LAURA VALLE

71 - MUTATIONS IN DNA POLYMERASE GENES (POLD1 & POLE) IN INDIVIDUALS HAVING EARLY-ONSET COLORECTAL CANCER AND/OR MULTIPLE ADENOMAS
GUY ROSNER

208 - MACROLIDE INDUCED READ-THROUGH OF APC NONSENSE MUTATIONS IN FAMILIAL ADENOMATOUS POLYPOSIS
RINA ROSIN-ARBESFELD

58 - MICRORNA EXPRESSION ASSOCIATED WITH DESMOID TUMOURS IN FAP
SARAH-JANE YVONNE WALTON

Review (20 minutes)
Cancer risk and *MUTYH* mutations
Stefan Aretz

**10:00 - Break (including electronic posters)**

**10:30 - Session 6**
Chairpersons
Susan Parry – Thomas Weber

**>Eldon Gardner Lecture** (20 minutes)
Introduction by Allan Spigelman

Management of upper GI tract in FAP
Andrew Latchford

**>Oral presentations** (60 minutes)
(presentation: 7 minutes + questions: 3 minutes)

199 - EXPERIENCE WITH PANCREAS-SPARING DUODENECTOMY FOR FAMILIAL ADENOMATOUS POLYPOSIS
R MATTHEW WALSH

24 - RISK MODIFYING FACTORS IN PATIENTS WITH PMS2 OR MUTYH MUTATIONS
SANNE WILLY TEN BROEKE

80 - EXPANDING THE MUTATION SPECTRUM AND PHENOTYPE OF POLYMERASE PROOFREADING-ASSOCIATED POLYPOSIS: NOVEL AND PREVIOUSLY REPORTED POLE VARIANTS IN AN ITALIAN SERIES
MAURIZIO GENUARDI

48 - EXPERIENCES AND ATTITUDES TOWARDS DIRECTLY APPROACHING INDIVIDUALS AT HIGH RISK OF HEREDITARY CANCER
HELLE VENDEL PETERSEN

39 - DUODENAL DISEASE IN MAP
SARAH-JANE YVONNE WALTON

151 - RANDOMIZED COMPARISON OF SURVEILLANCE INTERVALS IN FAMILIAL COLORECTAL CANCER
SIMONE DÉSIRÉE HENNINK
>**Debate – interactive** (20 minutes)
   Laparoscopic surgery and desmoid risk
   Yes - James Church vs. No - Lucio Bertario
   Moderated by Miguel Rodriguez-Bigas

**12:30 – Lunch (including electronic posters)**

**12:30-14:00**
InSiGHT Variant Interpretation Committee (VIC) Meeting

**14:00 - Session 7**
Chairpersons
Elena Stoffel – Miguel Rodriguez-Bigas

>**Dick Bussey Lecture** (20 minutes)
   Introduction by Gabriela Moeslein
   Risk reduction strategies for hereditary CRC
   John Burn

>**Oral presentations** (60 minutes)
   (presentation: 7 minutes + questions: 3 minutes)

150 - EXOME SEQUENCING IDENTIFIED POTENTIAL CAUSATIVE CANDIDATE GENES FOR HYPERPLASTIC POLYPOSIS SYNDROME
CHRISTINA ASTRID TRUECK

110 - LONG TERM DATA FOR CHEMOPREVENTION IN COLORECTAL DISEASE IN FAMILIAL ADENOMATOUS POLYPOSIS (FAP)
ANDREW LATCHFORD

200 - INCREASING INCIDENCE OF COLORECTAL CANCER (CRC) AMONG YOUNG ADULTS IN THE U.S. CHALLENGES INSIGHT AND CURRENT EPIDEMIOLOGIC TOOLS TO EXPLAIN AND REVERSE THE TREND
THOMAS KENNETH WEBER
123 - BETA2-MICROGLOBULIN MUTATIONS AND NK CELL MEDIATED CYTOTOXICITY IN MICROSATELLITE UNSTABLE COLORECTAL CANCER
MATTHIAS KLOOR

148 - MMR-DEFICIENT CRYPT FOCI AS CANCER PRECURSORS IN LYNCH SYNDROME – EVIDENCE FROM TUMOR HISTOLOGY
AYSEL AHADOVA

192 - FREQUENCY OF CDH1 GERMLINE MUTATIONS IN EARLY-ONSET GASTRIC CANCER IN BRAZIL
RODRIGO SANTA CRUZ GUINDALINI

>Review – Ileal Pouches
  Pouch for clinicians and surgeons – Gabriela Moeslein (10 minutes)
  The aging pouch – James Church (10 minutes)

16:00 - Break (including electronic posters)

16:00 – 17:00
InSiGHT Business Meeting

17:00 - Session 8 – Kay Neale Session
Chairpersons
Fabio Ferreira – Fabio Guilherme Campos

>Debate – Interactive (20 minutes)
  Does genotype influence surgical decision making in FAP and Lynch syndrome?
  Miguel Rodriguez-Bigas and Susan Clark
  Moderated by Gabriel Capellá

>David Jagelman Lecture (20 minutes)
  Introduction by Maurizio Genuardi

  From Leeds Castle Polyposis Group & International Collaborative Group in Hereditary Non-Polyposis Colorectal Cancer – ICG-HNPCC – to InSiGHT
  Patrick Lynch
78 - CAN ORAL REHYDRATION THERAPY CORRECT THE METABOLIC DISTURBANCES AND IMPROVE QUALITY OF LIFE AFTER COLECTOMY?
SREELAKSHMI MALLAPPA

176 - SURVIVAL RATE OF PATIENTS WHO DEVELOP CANCER IN RECTAL STUMP AFTER COLECTOMY AND IRA IN FAP PATIENTS
MARCO VITELLARO

77 - SURGICAL MANAGEMENT OF MYH-ASSOCIATED POLYPOSIS: IS MORE BETTER?
MATTHEW F KALADY

38 - URETERIC COMPLICATIONS OF INTRA-ABDOMINAL DESMOIDS
SARAH-JANE YVONNE WALTON

101 - UTILITY OF SINGLE NUCLEOTIDE POLYMORPHISMS TO GUIDE RISK APPROPRIATE COLORECTAL CANCER SCREENING
MARK JENKINS

124 - MOLECULAR ALTERATIONS IN MISMATCH REPAIR-DEFICIENT CRYPT FOCI IN LYNCH SYNDROME
MATTHIAS KLOOR

68 - ADENOMAS IN LYNCH SYNDROME: THE PERFECT STORM OF COLORECTAL CARCINOGENESIS
JAMES MICHAEL CHURCH

88 - MISS-RATE AND DELAY IN DIAGNOSIS OF SERRATED POLYPOSIS SYNDROME IN A CLINICAL COHORT
YASMIJN JOSANNE VAN HERWAARDEN

**18:30 – End of the day**

**20:30 - Dinner**
June 20th 2015 – Saturday

9:00 - Session 9
Chairpersons
Raul Cutait – Andrew Latchford

>TOP 3 Abstracts (30 minutes)
(presentation: 7 minutes + questions: 3 minutes)

11 - ENVIRONMENTAL MODIFIERS FOR THE RISK OF COLORECTAL CANCER IN LYNCH SYNDROME
AUNG KO WIN

96 - A PHASE 3 PLACEBO-CONTROLLED TRIAL OF CELECOXIB IN PEDIATRIC SUBJECTS WITH FAMILIAL ADENOMATOUS POLYPOSIS
CAROL BURKE

121 - VACCINATION OF MSI-H COLORECTAL CANCER PATIENTS WITH FRAMESHIFT PEPTIDE ANTIGENS – A PHASE I/IIA CLINICAL TRIAL
MATTHIAS KLOOR

>Jeremy Jass Lecture (20 minutes)
Introduction by Pål Møller

- Serrated Polyposis Syndromes
- Randall W. Burt

10:00 – Break (including electronic posters)

10:30 - Session 10
Chairpersons
Inge Bernstein – Susan Clark

>Review (20 minutes)
- PTEN Syndromes
- Brandie Leach

>Open Lecture (20 minutes)
Introduction by Luiz Fernando Lima Reis

- Genetic profile of the Brazilian population
>Round Table (10 minutes presentations - 70 minutes)
Global Collaborations: The way forward
Moderated by John Burn and James Church

107 - WORLDWIDE STUDY OF CANCER RISKS FOR LYNCH SYNDROME: INTERNATIONAL MISMATCH REPAIR CONSORTIUM (IMRC)
MARK JENKINS

15 - PROSPECTIVE CANCER RISKS AND SURVIVAL IN MMR MUTATION CARRIERS HAVING SURVIVED FIRST CANCERS
PÅL MØLLER

InSiGHT AND THE HUMAN VARIOME PROJECT: THE VARIANT INTERPRETATION COMMITTEE
MAURIZIO GENUARDI

COLON CANCER FAMILY REGISTER (CCFR)
FINLAY MACRAE

MALLORCA GROUP – WWW.MALLORCA-GROUP.EU
GABRIELA MOESLEIN

COLLABORATIVE GROUP OF THE AMERICAS – INHERITED COLORECTAL CANCER
CGA-ICC – WWW.CGA-ICC.ORG
ELENA STOFFEL

181 - GRUPO DE ESTUDIOS DE TUMORES HEREDITARIOS (GETH)
STUDY GROUP ON HEREDITARY TUMORS – WWW.GETH.ORG.BR
BENEDITO MAURO ROSSI

12:30 – Closing Remarks

Benedito Mauro Rossi

Invitation for the 7th. Biennial InSiGHT Meeting – Firenze – Italy
Maurizio Genuardi