

CURRICULUM VITAE

Name: Enrico Cappelli
Place and date of birth: GENOVA December 14th, 1964
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Personal history: September 1984-March 1986: National Service
April 1986-December 1992: management and selling activity in a tobacconist shop
July 4th, 1998: married with Anna Redaelli
May 4th, 2000: birth of Maddalena (daughter)
June 18th, 2003: birth of Giovanni (son)

Educational background

1978-1983: Agriculture Technical Institute G. Raineri, Piacenza. School-leaving certificate obtained on July 1983.
1987-1992: University of Genova. Degree in Biological Sciences obtained on November 1992. Thesis project: Effects of neutral Ca²⁺ ionophore on sea urchin block polyspermy and early development. Supervisor: Professor Carla Falugi.
March 1993 - March 1994: postdoctoral training at the Istituto Nazionale per la Ricerca sul Cancro, DNA Repair Unit (Dr. Guido Frosina).
April 1994: Qualifying examination as a Registered Biologist.
1993-1996: Post Doctoral School in Genetics, University of Pavia.

Professional record

December 1992 - March 1993: Fellow biologist at San Martino General Hospital - Hematology Department, Genova, Italia.

July 1994 - July 1996: postdoctoral fellowship at the Istituto Nazionale per la Ricerca sul Cancro, DNA Repair Unit (Dr. Guido Frosina), Genova, Italia. Studies on repair of damage induced by the alkylating antitumor drug chloroethylnitrosourea (CCNU) by an in vitro excision repair system.

August 1996 – June 2002: postdoctoral contract at the Istituto Nazionale per la Ricerca sul Cancro, DNA Repair Unit (Dr. Guido Frosina). Studies on DNA base excision repair mechanisms by an in vitro excision repair system.

July 2002 – August 2004: Marie Curie Fellowship, Medical Research Council, Radiation and Genome Stability Unit (RAGSU), DNA repair Group (Prof. John Thacker), Harwell, UK. Studies the role of recombination repair protein xrcc2 in correct chromosome segregation.

October 2004 – December 2005: postdoctoral contract at the Universitat Autònoma de Barcelona, Department of Genetic and Microbiology, Mutagenesis Laboratory (Dr. Jordi Surralles), Barcelona, Spain. Studies the DNA repair role of FANCD2 in different phase of the cell cycle.

February 2006 – Present: postdoctoral contract at the G. Gaslini Hospital, Department of Haematology, Haematology laboratory (Dr. C. Dufour), Genova. Complementation group analysis

of Italian Fanconi Anemia (FA) patient and studies on mechanisms leading to marrow failure in FA.

Publications (full papers):

- 1 - **Cappelli, E.**, Redaelli, A., Rivano, M.E., Abbondandolo, A., Frosina, G. (1995) Repair of 1-(2-chloroethyl)-3-cyclohexyl-1-nitrosourea-induced damage by mammalian cell extracts, *Carcinogenesis*, 16(9):2267-70.
- 2 - Falugi, C., Trielli, F., Germano, R., **Cappelli, E.** and Prestapino, G. (1995) Effects of neutral Ca²⁺ ionophore on sea urchin block polyspermy and early development, *Animal Biology*, 4: 51-58.
- 3 - **Cappelli, E.**, Taylor, R., Cevasco, M., Abbondandolo, A., Caldecott, K. and Frosina, G. (1997) Involvement of XRCC1 and DNA ligase III gene products in DNA base excision repair, *J. Biol. Chem.*, 272: 23970-23975.
- 4 - Taylor, R.M., Whitehouse, J., **Cappelli, E.**, Frosina, G. and Caldecott, K.W. (1998) Role of the DNA ligase III zinc finger in polynucleotide binding and ligation, *NAR*, 26: 4804-4810.
- 5 - Frosina, G., **Cappelli, E.**, Fortini, P. and Dogliotti, E. (1999) In vitro base excision repair assay using mammalian cell extracts, *Methods in Molecular Biology*, 113: 301-315.
- 6 - **Cappelli, E.**, Carrozzino, F., Abbondandolo, A. and Frosina, G. (1999) The DNA helicases acting in nucleotide excision repair XPD, CSB and XPB are not required for PCNA-dependent repair of abasic sites, *European J. Biochemistry*, , 259: 325-330.
- 7 - Rossi, O., Carrozzino, F., **Cappelli, E.**, Carli, F. and Frosina, G. (2000) Analysis of repair of abasic sites in early onset breast cancer patients, *Int. J. Cancer*, 85: 21-26.
- 8 - **Cappelli, E.**, Degan, P. and Frosina, G. (2000) Comparative repair of the endogenous lesions 8-oxo-7,8-dihydroguanine (8-oxoG), uracil and abasic site by mammalian cell extracts: 8-oxoG is poorly repaired by human cell extracts, *Carcinogenesis*, 21(6): 1135-1141.
- 9 - **Cappelli, E.**, Degan, P., Thompson, L. and Frosina, G. (2000) Efficient repair of 8-oxo-7, 8-dihydrodeoxyguanosine in Human and Hamster Xeroderma Pigmentosum D cells, *Biochemistry*, 39(34): 10408-10412.
- 10 - **Cappelli, E.**, Rossi, O., Chessa, L. and Frosina, G. (2000) Efficient DNA base excision repair in ataxia telangiectasia cells, *Eur. J. Biochem.*, 267: 6883-6887.
- 11 - **Cappelli, E.**, Hazra, T., Hill, J.W., Slupphaug, G., Bogliolo, M. and Frosina, G. (2001) Rates of base excision repair are not solely dependent on levels of initiating enzymes, *Carcinogenesis*, 22(3): 387-393.
- 12 - Bogliolo M, **Cappelli E**, D'Ostualdo A, Rossi O, Barbieri O, Kelley MR, Frosina G. (2002) Effect of *S. cerevisiae* APN1 protein on mammalian DNA base excision repair. *Anticancer Res.*, 22(5):2797-804.
- 13 - **Cappelli E**, D'Ostualdo A, Bogliolo M, Kelley MR, Frosina G. (2003) *Drosophila* S3 ribosomal protein accelerates repair of 8-oxoguanine performed by human and mouse cell extracts. *Environ Mol Mutagen.*, 42(1):50-8.

- 14 - **Cappelli E**, D'Osualdo A, Bogliolo M, Xu Y, Kelley MR, Frosina G. (2003) Repair of 8 oxoguanine in mammalian cells expressing the Drosophila S3 ribosomal/repair protein. *Teratog Carcinog Mutagen.*, Suppl 1:113-21.

- 15 - Alcalay M, Meani N, Gelmetti V, Fantozzi A, Fagioli M, Orleth A, Riganelli D, Sebastiani C, **Cappelli E**, Casciari C, Sciurpi MT, Mariano AR, Minardi SP, Luzi L, Muller H, Di Fiore PP, Frosina G, Pelicci PG. (2003) Acute myeloid leukemia fusion proteins deregulate genes involved in stem cell maintenance and DNA repair. *J Clin Invest.*, 112(11):1751-61.

- 16 - Frosina G., **Cappelli E**, Ropolo M., Fortini P., Pascucci B., Dogliotti E. (2006) In vitro base excision repair assay using mammalian cell extracts. *Methods Mol. Biol.*; 96: 314-377

- 17 - Bogliolo M., Lyakhovich A., Callén E., Castellà M., **Cappelli E.**, Ramírez M.J., Creus A., Marcos R., Kalb R., Neveling K., Schindler D. and Surrallés J. (2007) Histone H2AX and Fanconi anemia function in the same pathway to maintain chromosome stability. *Embo J.* 26(5):1340-51.

- 18 – M. Ropolo, A. Daga, F. Griffero, M. Foresta, G. Casartelli, A. Zunino, A. Poggi, **E. Cappelli**, G. Zona , R. Spaziante , G. Corte and G. Frosina (2009) A comparative analysis of DNA repair in stem and non-stem glioma cell cultures. *Mol. Cancer Res. Mol Cancer Res.* 7(3):383-92

- 19 – **E. Cappelli**; S. Townsend; C. Griffin and J. Thacker (2011) Homologous recombination proteins are associated with centrosomes and are required for mitotic stability. *Exp. Cell Res.* May 1;317(8):1203-13 **I.F. 2.58**

- 20 – M. Ropolo, **E. Cappelli**, M. Foresta, A. Poggi, L. Proietti-De-Santis and G. Frosina (2011) Defective resolution of pH2AX foci and enhanced DNA breakage in ionizing radiation-treated Cockayne syndrome B cells; *IUBMB Life* Apr;63(4):272-6 **I.F. 2.514**

- 21 – M. V. Corrias, R. Haupt, B. Carlini, **E. Cappelli**, S. Giardino, G. Tripodi, G.Tonini, A. Garaventa, V. Pistoia, A. Pistorio (2011) Multiple target molecular monitoring of bone marrow and peripheral blood samples from patients with localized neuroblastoma and healthy donors. *Pediatr Blood Cancer.* 2012 Jan;58(1):43-9. **I.F. 2.353**

- 22 – A. Raso, D. Vecchio, **E. Cappelli**, M. Ropolo, A. Poggi, P. Nozza, R. Biassoni, S. Mascelli, V. Capra, F. Kalfas, P. Severi and G. Frosina (2012) Specific sensitization of glioma stem cells to ionizing radiation by the ATM inhibitor KU-55933; *Brain Pathol.* 22(5):677-88 **I.F. 1.583**

- 23 – P. Anur, J. Yates, S. Vanderwerf, W. Keeble, K. Rathbun, M. Garbati, J.W. Tyner, J. Svahn, **E. Cappelli**, C. Dufour and G.C. Bagby. (2012) p38 MAPK inhibition suppresses the TLR-hypersensitive phenotype in FANCC- and FANCA-deficient mononuclear phagocytes; *Blood.* 119(9):1992-2002 **I.F. 9.06**

- 24 - **Cappelli E**, Vecchio D, Frosina G. (2012) Delayed formation of FancD2 foci in glioma stem cells treated with ionizing radiation. *J Cancer Res Clin Oncol.* 138(5):897-9 **I.F. 3.29**

- 25 - Korthof E., Svahn J., Terranova P., Peffault de Latour R., Soulier J., Socie G., Moins E., Pistorio A., Corsolini F., Fenoglio D., Dufour C. and **Cappelli E.** (2013) Immunological profile of Fanconi Anemia. A multicentric retrospective analysis of 61 patients; *Am J Hematol.* 88(6):472-6 **I.F. 3.477**

26) – Silvia Ravera; Daniele Vaccaro; Paola Cuccarolo; Marta Columbaro; Cristina Capanni; Martina Bartolucci; Isabella Panfoli; Alessandro Morelli; Carlo Dufour; **Enrico Cappelli** and Paolo Degan (2013) Mitochondrial respiratory chain complex I defects in Fanconi Anemia complementation group A; *Biochimie* 95(10):1828-37 **I.F. 3.123**

27 – C. Capanni, M. Bruschi, P. Cuccarolo, C., Ravera S., Dufour, G. Candiano, A. Petretto, P. Degan, **E. Cappelli**. (2013) Changes in vimentin, lamin A/C and mitofilin induce aberrant cell organization in fibroblasts from Fanconi anemia complementation group A (FA-A) patients.; *Biochimie*, 95(10):1838-47 **I.F. 3.123**

28 - **Cappelli E**, Ravera S, Vaccaro D, Cuccarolo P, Bartolucci M, Panfoli I, Dufour C, Degan P. (2013) Mitochondrial respiratory complex I defects in Fanconi anemia. *Trends Mol Med*. 19(9):513-4. **I.F. 10.11**

29 – Comar M, De Rocco D, **Cappelli E**, Zanotta N, Bottega R, Svahn J, Farruggia P, Misuraca A, Corsolini F, Dufour C, (2013) Fanconi Anemia Patients Are More Susceptible to Infection with Tumor Virus SV40. *PLoS One*. 8(11):e79683. **I.F. 3.534**

30 - De Rocco D, Bottega R, **Cappelli E**, Cavani S, Criscuolo M, Nicchia E, Corsolini F, Greco C, Borriello A, Svahn J, Pillon M, Mecucci C, Casazza G, Verzegnassi F, Cugno C, Locasciulli A, Farruggia P, Longoni D, Ramenghi U, Barberi W, Tucci F, Perrotta S, Grammatico P, Hanenberg H, Della Ragione F, Dufour C, and Savoia A. (2014) Molecular analysis of Fanconi anemia: the experience of the Bone Marrow Failure Study Group of the Italian Association of Pediatric Onco-Hematology. *Haematologica*.; 99(6):1022-31 **I.F. 5.814**

31 - Chiara Mattioli, Daniela De Rocco, Anna Monica Rosaria Bianco, Giulia Pianigiani, **Enrico Cappelli**, Anna Savoia, and Franco Pagani (2014) Unusual splice site mutations disrupt FANCA exon 8 definition; *BBA - Molecular Basis of Disease* 1842(7):1052-8 **I.F. 5.158**

32 - Guang-Hui Liu, Keiichiro Suzuki, Mo Li, Jing Qu, Nuria Montserrat, Carolina Tarantino, Ying Gu, Fei Yi, Xiuling Xu, Weiqi Zhang, Sergio Ruiz, Nongluk Plongthongkum, Kun Zhang, Shigeo Masuda, Emmanuel Nivet, Yuji Tsunekawa, Rupa Devi Soligalla, April Goebel, Emi Aizawa, Na Young Kim, Jessica Kim, Ilir Dubova, Ying Li, Ruotong Ren, Chris Benner, Antonio del Sol, Juan Bueren, Juan Pablo Trujillo, Jordi Surrallès, **Enrico Cappelli**, Carlo Dufour, Concepcion Rodriguez Esteban and Juan Carlos Izpisua Belmonte. Modeling Fanconi Anemia pathogenesis and therapeutics using integration-free patient iPSCs, *Nature Communication*, 2014 Jul 7;5:4330. **I.F. 11.47**

33 – Marta Columbaro, Silvia Ravera, Cristina Capanni, Isabella Panfoli, Giorgia Stroppiana, Paolo Degan, **Enrico Cappelli**. Treatment of FANCA cells with resveratrol and N-acetylcysteine: a comparative study, *Plos One* 2014 Aug 15;9(7):e104857. **I.F. 3.234**

34 - Silvia Ravera, Cristina Capanni, Danika Tognotti, Roberta Bottega, Marta Columbaro, Carlo Dufour, **Enrico Cappelli**, Paolo Degan. Inhibition of metalloproteinase activity in FANCA is linked to altered oxygen metabolism. *J Cell Physiol*. 2015 Mar;230(3):603-9. **I.F. 4.155**

- 35 – Johanna Svahn, Tiziana Lanza, Keaney Rathbun, Grover Bagby, Silvia Ravera, Fabio Corsolini, Angela Pistorio, Daniela Longoni, Piero Farruggia, Carlo Dufour, **Enrico Cappelli**; BIRB796, a p38MAPK inhibitor, stimulates in vitro hematopoietic progenitor and stem-cell colony growth in Fanconi Anemia group A patients, *Experimental Hematology*, 2015, 43(4):295-9 **I.F. 2.303**
- 36 - Cesare Usai, Silvia Ravera, Paola Cuccarolo, Isabella Panfoli, Carlo Dufour, **Enrico Cappelli**, Paolo Degan. Dysregulated Ca^{2+} homeostasis in Fanconi's anemia cells. *Scientific Reports* 2015, 5: 8088. **I.F. 5.228**
- 37 - Alessia Parodi, Francesca Kalli, Johanna Svahn, Giorgia Stroppiana, Daniela Derocco, Paola Terranova, Carlo Dufour, Daniela Fenoglio, **Enrico Cappelli**. Impaired immune response to *Candida albicans* in cells from Fanconi Anemia patients, *Cytokine* 2015, 73(1):203-7. **I.F. 2.94**
- 38 - Elena Nicchia, Francesco Benedicenti, Daniela De Rocco, Chiara Greco, Roberta Bottega, Francesca Inzana, Michela Faleschini, Serena Bonin, **Enrico Cappelli**, Massimo Moggi, Franco Stanzial, Johanna Svahn, Carlo Dufour, Anna Savoia. Clinical aspects of Fanconi anemia individuals with the same mutation of FANCF identified by next generation sequencing. *Birth Defects Research Part A: Clinical and Molecular Teratology*. 2015, doi: 10.1002/bdra.23388. **I.F. 1.954**
- 39 - Elena Nicchia, Chiara Greco, Daniela De Rocco, Vanna Pecile, Angela D'Eustacchio, **Enrico Cappelli**, Alberto Pallavicini, Lucio Torelli, Anna Savoia. Identification of point mutations and large intragenic deletions in Fanconi anemia using next generation sequencing technology, *Molecular Genetics & Genomic Medicine* 2015, doi: 10.1002/mgg3.160.
- 40 - Paolo Degan, Cesare Usai, Silvia Ravera, Paola Cuccarolo, Isabella Panfoli, Carlo Dufour, **Enrico Cappelli**; Altered Calcium and Red-ox homeostasis underline defective haematopoiesis in Fanconi Anemia, *Science Proceedings* 2015; 2: e880. doi: 10.14800/sp.880.
- 41 - Johanna Svahn, Francesca Bagnasco, **Enrico Cappelli**, Daniela Onofrillo, Silvia Caruso, Fabio Corsolini, Daniela De Rocco, Anna Savoia, Daniela Longoni, Marta Pillon, Aldo Misuraca / Nicoletta Marra, Ugo Ramenghi, Piero Farruggia, Anna Locasciulli, Carmen Addari, Carla Cerri / Elena Mastrodicasa, Gabriella Casazza, Federico Verzeegnassi, Angelica Barone, Simone Cesaro, Chiara Cugno, Giovanni Amendola, Carlo Dufour. Somatic, hematologic phenotype, long-term outcome and effect of hematopoietic stem cell transplantation. An analysis of 97 Fanconi Anemia patients from the Italian national database on behalf of the Marrow Failure Study Group of the AIEOP (Italian Association of Pediatric Hematology-Oncology), *Am J Hematol*. 2016, doi: 10.1002/ajh.24373. **I.F. 5.275**
- 42 - Silvia Ravera, Carlo Dufour, Simone Cesaro, Roberta Bottega, Michela Faleschini, Paola Cuccarolo, Fabio Corsolini, Cesare Usai, Marta Columbaro, Marco Cipolli, Anna Savoia, Paolo Degan and **Enrico Cappelli**; Evaluation of energy metabolism and calcium homeostasis in cells affected by Shwachman-Diamond syndrome, *Sci. Rep.* **6**, 25441; doi: 10.1038/srep25441 (2016). **I.F. 4.259**
- 43 - Paolo Degan, Silvia Ravera and **Enrico Cappelli** Why is an energy metabolic defect the common outcome in bone marrow failure syndromes? *Cell Cycle*, 2016, 11:1-5. **I.F. 3.530**

- 44 - **Enrico Cappelli**, Paola Cuccarolo, Giorgia Stroppiana, Maurizio Miano, Roberta Bottega, Vanessa Cossu, Paolo Degan, Silvia Ravera. Defect in mitochondrial energetic function compels Fanconi Anemia cells to glycolytic metabolism. *BBA - Molecular Basis of Disease* 2017, 1863(6):1214-1221. **I.F. 5.158**
- 45 - Emma Westermann-Clark, Alice Grossi, Francesca Fioredda, Stefano Giardino, **Enrico Cappelli**, Paola Terranova, Elena Palmisani, Jocelyn Farmer, Zsolt Foldvari, Yasuhiro Yamasaki, Maura Faraci, Edoardo Lanino, Luigi Notarangelo, Carlo Dufour, Isabella Ceccherini, Jolan E. Walter, Maurizio Miano RAG deficiency with ALPS features successfully treated with TCR $\alpha\beta$ /CD19 cell depleted haploidentical stem cell transplant; *Clinical Immunology*, 2018, 187:102-103. **I.F. 3.52**
- 46 - Roberta Bottega, Elena Nicchia, **Enrico Cappelli**, Silvia Ravera, Daniela De Rocco, Michela Faleschini, Fabio Corsolini, Filomena Pierri, Michaela Calvillo, Giovanna Russo, Gabriella Casazza, Ugo Ramenghi, Piero Farruggia, Carlo Dufour, Anna Savoia. Hypomorphic FANCA mutations correlate with mild mitochondrial and clinical phenotype in Fanconi anemia; *Haematologica*, 2018 Mar;103(3):417-426. **I.F. 7.702**
- 47 - Silvia Ravera, Vanessa Cossu, Barbara Tappino, Elena Nicchia, Carlo Dufour, Simona Cavani, Andrea Sciutto, Claudia Bolognesi, Marta Columbaro, Paolo Degan, **Enrico Cappelli**. Dose-dependent metabolic effects of metformin in healthy and Fanconi Anemia lymphoblast cells; *Journal cellular physiology*, 2018, 233(2):1736-1751. **I.F. 4.08**
- 48 - Silvia Ravera, Carlo Dufour, Paolo Degan, **Enrico Cappelli**. Fanconi anemia: from DNA repair to metabolism, *EJHG*, 2018 doi:10.1038/s41431-017-0046-6. **I.F. 4.287**
- 49 - Silvia Ravera, Paolo Degan, Federica Sabatini, Marta Columbaro, Carlo Dufour, **Enrico Cappelli**. Altered lipid metabolism could drive the bone marrow failure in Fanconi Anemia, *BJH*, 2018 DOI: 10.1111/bjh.15171. **I.F. 5.67**
- 50 - Maurizio Miano, Gioacchino Andrea Rotulo, Elena Palmisani, Mariateresa Giaimo, Francesca Fioredda, Filomena Pierri, Agnese Pezzulla, Maria Licciardello, Paola Terranova, Tiziana Lanza, **Enrico Cappelli**, Rosario Maggiore, Michaela Calvillo, Concetta Micalizzi, Giovanna Russo, Carlo Dufour, Sirolimus as a rescue therapy in children with immune thrombocytopenia refractory to mycophenolate mofetil (2018) *American Journal of Hematology* Accepted **I.F. 5.275**
- 51 - Paolo Degan, **Enrico Cappelli**, Maria Grazia Longobardi, Alessandra Pulliero, Paola Cuccarolo, Carlo Dufour, Silvia Ravera, Daniela Calzia, Alberto Izzotti. A global profile of microRNAs in Fanconi anemia; submitted *JCCS*
- 52 - Silvia Ravera, Paolo Degan, Fabio Corsolini, Barbara Banelli, Carlo Dufour, **Enrico Cappelli**. Are lipid metabolism, oxidative stress and insulin resistance correlated aspects of mitochondria disorder in Fanconi anemia cells? Submitted *JCP*

Genova, 16 March 2018

