Ottava Giornata della Ricerca della Svizzera Italiana
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Modulo per la sottomissione abstract ricerca di LABORATORIO

Titolo (massimo **15 parole**)

CIRCULATING TUMOR DNA REVEALS GENETICS, CLONAL EVOLUTION AND RESIDUAL DISEASE IN CLASSICAL HODGKIN LYMPHOMA

Autori (cognome e iniziali, es: Grassi L.)

Spina V.1*, Bruscaggin A.1*, Cuccaro A.2, Martini M.3, Di Trani M.4, Forestieri G.1, Manzoni M.5, Condoluci A.1,6, Arribas A.1, Terzi-Di-Bergamo L.,1 Locatelli SL.4, Cupelli E.2, Ceriani L.6, Moccia AA.6, Stathis A.6, Nassi L.7, Deambrogi C.7, Diop F.7, Guidetti F.1, Annunziata S.8, Rufini V.,8 Giordano A.8, Neri A.5,9, Gerber B.6, Bertoni F.1,6, Ghielmini M.6, Stüssi G.6, Santoro A.4, Cavalli F.1,6, Zucca E.6, Larocca LM.3, Gaidano G.7, Hohaus S.2^, Carlo-Stella C.4^, Rossi D.1,6^ *V.S. and A.B. equally contributed ^S.H., C.C.S. and D.R. equally

Affiliazioni (ospedale o istituto, servizio o reparto, indirizzo, es: Ospedale Regionale di Lugano, Servizio di angiologia, Lugano)

1Institute of Oncology Research, Bellinzona, Switzerland; 2Institute of Hematology, Policlinico Gemelli Foundation, Catholic University of the Sacred Heart, Rome, Italy; 3Division of Pathology, Policlinico Gemelli Foundation, Catholic University of the Sacred Heart, Rome, Italy; 4Humanitas Cancer Center, Humanitas University, Milan, Italy; 5Department of Oncology and Hemato-oncology, University of Milano, Milan Italy; 6Oncology Institute of Southern Switzerland, Bellin-zona, Switzerland; 7Division of Hematology, Department of

Testo (massimo **250 parole**, preferibilmente in italiano (accettato anche in inglese), suddiviso in Introduzione, ***Metodi***, ***Risultati***, ***Conclusioni*** e ***Finanziamento***

INTRODUCTION: The rarity of neoplastic cells in the biopsy imposes major technical hurdles that have so far limited genomic studies in classical Hodgkin lymphoma (cHL). By using a highly sensitive and robust deep-next-generation-sequencing approach for circulating tumor DNA (ctDNA), here we aimed at tracking the genetics of cHL in different clinical phases, and its modifications upon treatment. METHODS: The analysis was based on specimens collected from 80 newly diagnosed and 32 refractory cHL patients, including longitudinal samples collected under ABVD chemotherapy and longitudinal samples from relapsing patients treated with chemotherapy and immunotherapy. RESULTS: Overall, 87.5% of mutations discovered in cfDNA were identified in tumor genomic DNA, thus indicating that ctDNA mirrors the genotype of cHL. By genotyping on ctDNA a large cohort of newly diagnosed cHL, STAT6 (37.5%), TNFAIP3 (35.0%), and ITPKB (27.5%) turned out to be the most recurrently affected genes in this tumor. Affected genes pointed to the molecular deregulation of specific programs in newly diagnosed cHL, including, NF-κB (46.2%), PI3K/AKT (46.2%), and cytokines signaling (37.5%). Longitudinal ctDNA profiling identified treatment-dependent patterns of clonal evolution in patients relapsing after chemotherapy and patients maintained in partial remission under immunotherapy. By measuring ctDNA changes during therapy, we propose ctDNA as a radiation-free tool to track residual disease that may integrate PET imaging for the early identification of chemorefractory cHL patients. CONCLUSIONS: Collectively, our results provide the proof of concept that ctDNA may serve as novel precision medicine biomarker in cHL.

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Visto superiore (prego indicare Nome e Cognome del superiore)

Davide Rossi

Criteri per sottomissione Abstract:
NO Case report
NO Abstract senza nessun risultato
VISTO da un superiore

Invio Abstract