

Al Consiglio Nazionale delle Ricerche
Ufficio Accordi e Relazioni Internazionali
P.le Aldo Moro, 7
00185 Roma

Programma Short Term Mobility 2013 – Relazione dell'attività scientifica svolta

Dipartimento di appartenenza / Candidate's department : Scienze biomediche

Titolo del programma / Title : Identification of genomic regions of exceptionally high or low population differentiation from the 1000 Genomes Phase I data

Il fruitore/ Candidate: Vincenza Colonna

Istituto di appartenenza / Research Institute: Istituto di Genetica e Biofisica "Adriano Buzzati-Traverso" Qualifica / Position: Ricercatore (n. matricola 11803) livello / level terzo

Istituzione ospitante / Description of the host institution: Human evolution team - The Wellcome Trust Sanger Institute, Wellcome Trust Genome Campus Hinxton, Cambridgeshire CB10 1SA United Kingdom

The purpose of my visit to Sanger was to follow up a project aimed to the analyses of genetic differentiation human populations. During my visit to the Sanger Institute I accomplished the objective described in the program, applying the analyses indicated on it.

We identified regions of extreme population differentiation using insertion/deletions and structural variants demonstrating that those regions are subject to the effect of natural selection. In particular we identified high frequency of a human-specific mutation (ie not present in our closest relative) in gene related to skin phenotypes whose expression is regulated by the ultra violet radiation and thus possibly implicated in the process of skin adaptation in humans that migrated out of Africa 60,000 years ago.

We summarized results and we are in the stage to submit it for publication.

Napoli 01/07/2013

Vincenza Colonna

