GENOMICS AND EVOLUTION OF HUMAN PHENOTYPIC TRAITS

PhD Student: Virginia Veltre / *Supervisor*: Prof. Olga Rickards / *Co-supervisor*: Dr. Flavio De Angelis

34th Cycle - A.Y. 2018/2019

This research project aims to investigate the genomics involvement in the onset of several human phenotypic traits. Phenotypic variation is shaped through complex interactions between genotype and environment, and mounting evidence coming from GWAS studies suggest human genome variability could account for the differential expression of physical characteristics. Thus the molecular typing of the whole genome could ultimately result in the capability to develop inferring models able to quantify and predict multifactorial and complex traits such as facial shape, pigmentary traits of eyes, skin and hair and in addition ear morphology, body structure or male baldness. Furthermore, the whole genome sequencing allows for achieving information about the biogeographical origin of the samples that are useful to compile an integrated Individual Biological Profile.

Combining genomic and phenomic data, new bioinformatic and statistical tools will permit evaluation of the relative influences of genetic and environmental factors in determining the onset of specific characters that should be linked to potential constraints to phenotypic plasticity, that has long been recognized as a key strategy enabling organisms to respond to varying environments both adaptively and non-adaptively.

Several studies demonstrated that environmental heterogeneity favors plasticity but there is also the potential for variation in local environments to destabilize population homeostasis and consequently disrupt the match between an individual phenotype and environment. The reliability of environmental cues is of critical importance for plasticity to be favored within and across generations.

The genomic evaluation will be carried out on a whole sample of ca 200 samples pertaining to 10 different populations scattered in heterogeneous environmental landscapes. The information gained by extant population genomics will be complemented by a diachronic sample of 30 individuals belonging to different ancient populations to investigate the evolution of human phenotypic traits starting from the past related data. Next Generation Sequencing techniques will be employed to shotgun analyze the ancient and extant DNA as well as a target enrichment methodology will be devoted to achieve significant coverage for selected genomic markers able to proficiently allow the identification of putative associations between markers and phenotypic traits. This work paves the way to provide novel information by the identification of new evolutionary potential of human phenomic-related genomic characters. This approach offers the opportunity to shed new light on the constraints on phenotypic plasticity evolution in humans that could ultimately be a terrific asset also for personal identification useful in forensics to reach a reliable phenotypic identikit.

CONTRIBUTED PRESENTATIONS

2018

Veltre Virginia, De Angelis Flavio, Biondi Gianfranco, Rickards Olga Evaluation of skin-related variants in African ancestry populations and their role in personal identification. Presented at 7th Sardinian International Summer School "From GWAS to Function", Polaris Technology Park, Pula (CA), Italy. Awarded to be presented as oral presentation.

Veltre Virginia, Parisi Arianna, De Angelis Flavio, Biondi Gianfranco, Rickards Olga. Evaluation of skin-related variants in African ancestry populations and their role in personal identification. Presented at 87th AAPA Annual Meeting. Austin, TX (USA). Poster presentation.

2017

Veltre Virginia, Parisi Arianna, De Angelis Flavio, Biondi Gianfranco, Rickards Olga. Varianti correlate alla pigmentazione cutanea in popolazioni di origine africana: ruolo nell'identificazione personale. Presented at XXII Congress Associazione Antropologica Italiana. Rome, Italy. Poster presentation.

Parisi Arianna, Veltre Virginia, De Angelis Flavio, Carreri Silvia, Piccioli Andrea, Maccauro Giuseppe, Rickards Olga. Studio pilota sull'eziologia genetica del Sarcoma Osteogenico: valutazione di varianti causative in un campione di casi italiani. Presented at XXII Congress Associazione Antropologica Italiana. Rome, Italy. Poster presentation.

Veltre Virginia, Parisi Arianna, De Angelis Flavio, Biondi Gianfranco, Rickards Olga. Genetic contribution to human skin pigmentation in selected African ancestry populations. Presented at 10th ISABS Congress. Dubrovnik, Croatia. Poster presentation.

Parisi Arianna, Veltre Virginia, De Angelis Flavio, Piccioli Andrea, Rickards Olga, Maccauro Giuseppe, Carreri Silvia. Risk-associated SNPs in osteosarcoma: a pilot study to dissect its genetic heterogeneity. Presented at 10th ISABS Congress. Dubrovnik, Croatia. Poster presentation.

PUBBLICATIONS

Veltre Virginia, Parisi Arianna, De Angelis Flavio, Biondi Gianfranco, Rickards Olga. Evaluation of skin-related variants in African ancestry populations and their role in personal identification. American Journal of Physical Anthropology. 2018, 165 (S66) 287-288.