



EUROPEAN SOCIETY OF HUMAN GENETICS

ESHG 2015

Glasgow

Glasgow, Scotland, UK, June 6-9

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Analyses of the molecular pathways involved in the sarcopenic process in functional and non-functional elderly population.

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Abstract:

Abstract

Sarcopenia is one of the most deleterious effects of aging. The involuntary loss of muscle mass, strength, and muscular function have a major impact on quality of life in the elderly population. The etiology of the sarcopenia is not clearly established, although, a multifactorial process that develops from the fourth decade of life is proposed. In a previous clinical report we found that evaluation of the muscular contractility is a *per se* and independent factor in the sarcopenia development not related with the muscle volume. In order to understand this phenomenon, we analyzed the molecular pathways involved in the muscle performance and the sarcopenic process. Genetic expression patterns in functional and non-functional elderly groups were studied in 15 muscle biopsies from both groups, using a QRT-PCR array (i2Profiler, Qiagen). We studied critical signaling pathways involved in the sarcopenic process (apoptosis, autophagy, WNT, TGF-Beta, TNF-Alpha and interleukins). Immunohistochemistry and confocal analysis were also performed in order to explore differences in the distribution of the muscular proteins involved in the contractile process (actin, myosin, myopalladin and SERCA). Preliminary results showed an overexpression of genes involved in apoptosis, autophagy and interleukins in the non functional group. Confocal analysis showed a different protein distribution in a qualitative and quantitative fashion in both groups. Our data suggests that inflammatory and apoptotic process in the muscular fiber impacts the proteins involved in contractility and muscular functionality, observed in the clinic as less contractility, strength and muscular performance in the sarcopenic and non functional population.
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Topic (Complete): 04: Skeletal, connective tissue, ectodermal and skin disorders

Keyword (Complete): sarcopenia ; apoptosis ; molecular pathways

Presentation Preference (Complete): Oral preferred

Awards - Fellowships (Complete):



Session Overview

Plenary Sessions

Opening - Welcome Addresses	Saturday, June 6 14.00 - 14.30 hrs
P1. Opening Plenary Session	Saturday, June 6 14.30 - 16.00 hrs
P2. What's New?	Saturday, June 6 16.30 - 20.00 hrs
P3. Interactive Debate - Should all geneticists have their genome sequenced?	Tuesday, June 9 9.00 - 10.30 hrs
P4. Mendel Lecture	Tuesday, June 9 13.30 - 14.15 hrs
P5. ESHG Award Lecture, EHG Nature Awards, Young Investigator & Poster Awards, Closing	Tuesday, June 9 14.15 - 15.45 hrs

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Concurrent Symposia

S01. Big Data Genomics and Human Knock-outs	Sunday, June 7 8.30 - 10.00 hrs
S2. Building Bridges Symposium - Genetic testing in Children	Sunday, June 7 8.30 - 10.00 hrs
S03. Epigenetic Basis of Disease	Sunday, June 7 8.30 - 10.00 hrs
S04. Spliceosomeopathies	Sunday, June 7 8.30 - 10.00 hrs
S05. Reproductive genetics and "chromosome therapy"	Sunday, June 7 17.30 - 19.00 hrs
S06. International data sharing initiatives	Sunday, June 7 17.30 - 19.00 hrs
S07. Telomeres in Human Disease	Sunday, June 7 17.30 - 19.00 hrs
S08. Mouse Phenotyping for clinical research	Sunday, June 7 17.30 - 19.00 hrs
S09. Evolution of the human genome: Clinical implications	Monday, June 8

S10. From rare to common variants in cardiovascular diseases (joint with ESC)

Monday, June 8
8.30 - 10.00 hrs

S11. Non-coding DNA and human disease

Monday, June 8
8.30 - 10.00 hrs

S12. Mitochondria and Genetic Disease

Monday, June 8
8.30 - 10.00 hrs

S13. Therapeutic Strategies for Genetic Diseases

Monday, June 8
17.30 - 19.00 hrs

S14. Somatic Mutation Detection and Interpretation

Monday, June 8
17.30 - 19.00 hrs

S15. Evolution and Disease

Monday, June 8
17.30 - 19.00 hrs

S16. Genome Regulation

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17.30 - 19.00 hrs[To top](#)**Educational Sessions**

ES1. Translational Cancer Genetics

Saturday, June 6
10.30 - 12.00 hrs

ES2. CRISPR-Cas9

Saturday, June 6
10.30 - 12.00 hrs

ES3. Care for Rare Diseases

Saturday, June 6
10.30 - 12.00 hrs

ES4. From Genes to Networks

Saturday, June 6
10.30 - 12.00 hrs

EF Automating Clinical Genetics

Sunday, June 7
8.30 - 10.00 hrs

ES6. My vision on Genomic medicine

Sunday, June 7
11.45 - 13.15 hrs

ES7. Imprinting-related Disorders

Sunday, June 7
17.30 - 19.00 hrs

ES8. Palliative Care of Genetic Conditions

Monday, June 8
8.30 - 10.00 hrs

ES9. Mutation Prediction Tools

Monday, June 8
17.30 - 19.00 hrs[To top](#)**Concurrent Sessions**

C01. NIPT

Saturday, June 6
18.30 - 20.00 hrs

C02. Improvement in genome sequencing and functional studies	Saturday, June 6 18.30 - 20.00 hrs
C03. Novel genes causing intellectual disability	Saturday, June 6 18.30 - 20.00 hrs
C04. The many faces of cancer mutations	Saturday, June 6 18.30 - 20.00 hrs
C05. Cardiovascular disorders	Saturday, June 6 18.30 - 20.00 hrs
C06. Neuromuscular disorders	Saturday, June 6 18.30 - 20.00 hrs
C07. Reproductive Genetics	Sunday, June 7 13.15 - 14.45 hrs
C08. Integrative OMICS approaches in common traits	Sunday, June 7 13.15 - 14.45 hrs
C9. Genetic susceptibility to cancer development	Sunday, June 7 13.15 - 14.45 hrs
C10. Neurogenetic disorders	Sunday, June 7 13.15 - 14.45 hrs
C11. Skeletal disorders	Sunday, June 7 13.15 - 14.45 hrs
C12. Sensory disorders	Sunday, June 7 13.15 - 14.45 hrs
C13. Fundamental insights in structural genomics	Monday, June 8 13.15 - 14.45 hrs
C14. Challenges in genetic counselling	Monday, June 8 13.15 - 14.45 hrs
C15. Network and functional analysis in intellectual disability	Monday, June 8 13.15 - 14.45 hrs
C16. Growth failure and microcephaly	Monday, June 8 13.15 - 14.45 hrs
C17. Epigenetic control of gene expression	Monday, June 8 13.15 - 14.45 hrs
C18. Metabolic and renal disorders	Monday, June 8 13.15 - 14.45 hrs
C19. Diagnostic NGS	Tuesday, June 9 11.00 - 12.30 hrs
C20. Current issues in genet(h)ics	Tuesday, June 9 11.00 - 12.30 hrs
C21. Multiple congenital anomaly syndromes	Tuesday, June 9 11.00 - 12.30 hrs
C22. Statistical genetics and bioinformatics	Tuesday, June 9 11.00 - 12.30 hrs
C23. Movement and motor disorders	Tuesday, June 9

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Workshops

WS01. A case that changed my life as a clinical geneticists (TEDEX format)	Saturday, June 6 10.30 - 12.00 hrs
WS02. NGS Variant Analysis with Galaxy	Saturday, June 6 10.30 - 12.00 hrs
WS03. NGS in clinics	Sunday, June 7 15.30 - 17.00 hrs
WS04. Dysmorphology 1	Sunday, June 7 15.30 - 17.00 hrs
WS05. Pre-conception carrier testing	Sunday, June 7 15.30 - 17.00 hrs
WS06. Blurred boundaries between clinic and research	Sunday, June 7 15.30 - 17.00 hrs
WS08. Ensembl Highlights: What's New in Accessing our Genomes?	Sunday, June 7 15.30 - 17.00 hrs
WS08. European Board of Medical Genetics: What can we do to facilitate you to become a registered genetic professional?	Sunday, June 7 15.30 - 17.00 hrs
WS10. Practical Bioinformatics Whole exome sequence analysis	Monday, June 8 15.30 - 17.00 hrs
WS11. Dysmorphology 2	Monday, June 8 15.30 - 17.00 hrs
WS12. Reproductive genetics	Monday, June 8 15.30 - 17.00 hrs
WS13. Clinical Cancer Genetics	Monday, June 8 15.30 - 17.00 hrs
WS14. Copy Number Variant Interpretation and Classification	Monday, June 8 15.30 - 17.00 hrs
WS15. Genome Browser UCSC	Monday, June 8 15.30 - 17.00 hrs
WS16. Genetics Clinic of the Future	Monday, June 8 15.30 - 17.00 hrs

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