SYMPOSIUM
'CARE AND RESEARCH IN VASCULAR EHLERS-DANLOS SYNDROME AND HEREDITARY AORTOPATHIES: AN ITALIAN PERSPECTIVE'

Rome, Italy, Saturday 7th December 2024
Aula Magna, Policlinico Tor Vergata
Viale Oxford 81, 00133 Roma

Organizing Committee
Fabio Bertoldo, Fondazione Policlinico Tor Vergata, Rome, Italy
Marco Castori, Fondazione IRCCS-Casa Sollievo della Sofferenza, San Giovanni Rotondo, Italy
Pietro Gaudenzi, 'With Giacomo against Ehlers-Danlos vascolare syndrome', Rome, Italy
Maria Chiara Tealdo, 'With Giacomo against Ehlers-Danlos vascolare syndrome', Rome, Italy

Sponsorship
*XXX (**to be confirmed**)*

Speakers and Chairmen
Martina Berteotti, University of Florence and AOU Careggi, Florence, Italy
Fabio Bertoldo, Fondazione Policlinico Tor Vergata, Rome, Italy
Marco Castori, Fondazione IRCCS-Casa Sollievo della Sofferenza, San Giovanni Rotondo, Italy
Nicola Chiarelli, Brescia, Italy
Filippo Gaudenzi, Radio Televisione Italiana (RAI), Rome, Italy
Pietro Gaudenzi, 'With Giacomo contro Ehlers-Danlos vascolare syndrome', Rome, Italy
Betti Giusti, University of Florence and AOU Careggi, Florence, Italy
Lucia Micale, Fondazione IRCCS-Casa Sollievo della Sofferenza, San Giovanni Rotondo, Italy
Silvia Morlino, Fondazione IRCCS-Casa Sollievo della Sofferenza, San Giovanni Rotondo, Italy
Giuseppe Novelli, Tor Vergata University and Fondazione Policlinico Tor Vergata, Rome, Italy
Augusto Orlandi, Tor Vergata University and Fondazione Policlinico Tor Vergata, Rome, Italy
Marco Ritelli, University of Brescia, Brescia, Italy
*Annalisa Scopinaro, UNIAMO **(to be confirmed)***
Maria Chiara Tealdo, 'With Giacomo against Ehlers-Danlos vascolare syndrome', Rome, Italy

PATIENTS' ASSOCIATION
'WITH GIACOMO AGAINST EHLERS-DANLOS VASCULAR SYNDROME'

Dear Participants,

In November 2021, Giacomo's family established the Patients' Association 'With Giacomo against Ehlers-Danlos vascular syndrome', whose Memorandum, Articles, and financial statements are on file at RUNTS.

The decision to create the Association was determined by the hospitalisation of our son Giacomo at the San Giovanni Hospital in Rome, which lasted from May to October 2021, with multiple inpatients in the Intensive Care Unit. During this period, Giacomo was treated urgently for several aneurysms and an intestinal perforation caused by the lack of collagen resulting from vascular Ehlers-Danlos syndrome (vEDS).

Restarting research on this rare syndrome, which has been at a standstill in Italy for some time, is the main goal of the Association.

In the past three years, we have been pursuing the following goals:

- *To promote and support research activities and projects aimed at implementing knowledge for the diagnosis and treatment of vEDS and fostering a network of researchers;*
- *To raise awareness and inform the public on issues relevant to its aims and to promote greater knowledge of vEDS;*
- *To urge political authorities to take appropriate measures to improve the prevention, diagnosis and treatment of vEDS;*
- *To promote, at national and international level, the exchange of information and data;*
- *To promote and encourage the formation of a network between families affected by vEDS that enables mutual support and the exchange of experiences in the management of daily life in order to improve the quality of life of people affected by the disease;*
- *To maintain close relations with national and international organisations and associations that pursue the same goals.*

We can affirm that the Symposium entitled 'CARE AND RESEARCH IN VASCULAR EHLERS-DANLOS SYNDROME AND HEREDITARY AORTOPATHIES: AN ITALIAN PERSPECTIVE' is the ideal venue for the convergence of these aims and offers concrete opportunities to achieve them.

Maria Chiara Tealdo and Pietro Gaudenzi
Giacomo's parents

SYMPOSIUM PRESENTATION

The symposium 'CARE AND RESEARCH IN VASCULAR EHLERS-DANLOS SYNDROME AND HEREDITARY AORTOPATHIES: AN ITALIAN PERSPECTIVE' to be held on Saturday 7th December 2024 at the Tor Vergata University in Rome (Italy) represents a 'unicum' in the Italian medical-scientific panorama on hereditary connective tissue disorders and cardiogenetics.

Vascular Ehlers-Danlos syndrome is a prototype of hereditary vasculopathy in both nosological and clinical terms. This condition is determined by deleterious variants in the COL3A1 gene, which codes for the 'alpha' subunit of type III collagen. Similarly to the other variants of Ehlers-Danlos syndrome, the gene involved in the vascular form plays a crucial role in the morpho-functional integrity of the connective tissue.

Vascular Ehlers-Danlos syndrome is a very specific clinical variant of Ehlers-Danlos syndrome and its clinical picture is dominated by cardiovascular involvement. In particular, the life expectancy of affected inidviduals is compromised by the risk of arterial ruptures either spontaneously or during trauma and surgical procedures. Such phenomena commonly involve medium-size arteries and the intrathoracic aorta.

On a clinical perspective, vascular Ehlers-Danlos syndrome shows similarities with other inherited connective tissue disorders and hereditary vasculopathies, such as Loeys-Dietz syndrome, Marfan syndrome, hereditary haemorrhagic telangiectasia and familial thoracic aortic aneurysm.

These similarities appear relevant for diagnostic and management issues, and stimulate the medical community to converge expertise and activities in ultra-specialised centres. Moreover, they appear as an opportunity for the development of innovative therapies that may be of benefit to people suffering from molecularly distinct but clinically overlapping conditions.

This Symposium aims to focus on the basic and pre-clinical research on vascular Ehlers-Danlos syndrome carried out in Italy, in order to facilitate the transition towards clinical and translational research.

Fabio Bertoldo and Marco Castori

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DETAILED PROGRAM

09.30 - 10.00 Opening of the Symposium and Opening Remarks (Fabio Bertoldo, Maria Chiara Tealdo, Pietro Gaudenzi)

10.00-10.30 ***LECTURE:*** *Contribution of the OMIC sciences to cardiovascular medicine (Giuseppe Novelli)*

PRINCIPLES OF TAILORED MEDICINE IN PEOPLE WITH HEREDITARY AORTOPATHY/ARTERIOPATHY
(Chairmen: Filippo Gaudenzi, Fabio Bertoldo)

PART ONE
10.30-10.50 Current molecular diagnostics in Cardiogenetics: How and Why (Marco Castori)
10.50-11.10 Precision medicine in Cardiogenetics: patients' pathways (Martina Berteotti)

11:10 - 11:30 Coffee Break

PART TWO
11.30-11.50 Precision medicine in Cardiogenetics: drug therapy (Silvia Morlino)
11.50-12.10 Precision medicine in Cardiogenetics: surgical procedures (Fabio Bertoldo)
12.10-12.40 Q&A

12:40 - 14:00 Light Lunch

14.00-14.30 The Telethon Foundation's commitment to research on rare diseases ***(to be defined)***

PRE-CLINICAL RESEARCH AND NOVEL THERAPEUTIC PERSPECTIVES
(Chairmen: Filippo Gaudenzi, Annalisa Scopinaro, Betti Giusti)

PART ONE
14.30-14.50 Therapeutic innovations in vascular Ehlers-Danlos syndrome: basic, pre-clinical and clinical research (Nicola Chiarelli)
14.50-15.10 Pre-clinical research and therapeutic perspectives for hereditary aortopathies and arteriopathies in Northern Italy: University of Brescia (Marco Ritelli)

15:10 - 15:30 Coffee Break

PART TWO
15.30-15.50 Pre-clinical research and therapeutic perspectives for hereditary aortopathies and arteriopathies in Central Italy: Tor Vergata University (Augusto Orlandi)
15.50-16.10 Pre-clinical research and therapeutic perspectives for hereditary aortopathies and arteriopathies in Southern Italy: Fondazione Casa Sollievo della Sofferenza (Lucia Micale)
16.10-16.40 Q&A

16.40 Closure of the Symposium and Future Perspectives (Fabio Bertoldo, Maria Chiara Tealdo, Pietro Gaudenzi)