



2017 Montalcino Aortic Consortium (MAC) Summit

**Borgo San Luigi, Monteriggioni (SI), Italy.
May 30 – June 2, 2017**

AGENDA:

Tuesday, May 30, 2017

Arrivals, 17:30 – Welcome reception
19:00 – Dinner

Wednesday, May 31, 2017

8:00 – 12:00

Welcome and Introductions

I. Montalcino Aortic Consortium: State of the Consortium

A. Review of MAC and relevant related studies:

TGFBR1/TGFBR2

SMAD, MYLK, and Classification of Aortopathies

ClinGen

Outcomes Based on Recruitment Sites

Guillaume

Dianna

Julie

Ellen

B. MAC infrastructure and database and other registries

MAC

VASCERN

GenTAC

STS Adult Cardiac Surgery Database

Marfan Foundation Patient Registry

IRAD

SPR

Dianna

Guillaume

Reed

Joe

Jo

Arturo

Shaine

C. Funding for MAC

Dianna and Guillaume

Pending or Planned grant applications:

1. **Marfan Foundation:** Use MAC infrastructure to characterize and optimized care for neonatal MFS.
2. **NIH Notice of Intent: NOT-HL-17-491** U01 Investigator –initiated epidemiological cohort studies. Funding for continued surveillance for outcomes, maintain biospecimens and database and exams. Establish a platform to support innovative science through ancillary studies funded through other research support.
3. **US Diagnostic Companies**

D. Translating new knowledge into improved diagnoses and clinical management

Reed, Guillaume, Dianna

Post data with or without recommendations; “Get with the Data”

Websites: GADA, JRF, MF,

YouTube videos

Webcasts: John Ritter Foundation and Houston experience

12:00 – 13:00 – Lunch

13:00 – 15:30

II. MAC Research Opportunities

Shaine

Methods for retrospective and prospective clinical studies

Laura and Dianna

ACMG Variant Classification for FBN1 and other HTAD genes

Julie

Mitral Valve Disease for HTAD

Myocardial dysfunction associated with HTAD

Guillaume and Marjolijn

Pregnancy Associated Risk for Dissection

Guillaume and Checco

Medical Therapy for HTAD

Guillaume and Joe

Surgical Outcomes in HTAD cases

Ellen

Facial features associated with HTAD genes

Sherene (Dianna)

Medical and surgical management of descending aortic dissections in HTAD cases

16:00 Montalcino & Dinner

Thursday, June 1, 2017

8:00 – 12:00

III. MAC Research Opportunities

Dianna

Modifiers of *ACTA2* R149C Phenotype

Neonatal Marfan syndrome

Ellen and Guillaume

Aortic phenotype for *TGF β 2*, *PRKG1*, *LOX*, *TGF β 3*, other HTAD genes

TGF β 1 vascular disease gender differences

Shaine

Vertebral Artery Tortuosity as a Biomarker for Dissection

FLNA Mutations: CV phenotype

Siddharth

BAV phenotype associated with HTAD gene
Morphology of the SV and aortic arch and risk for dissection

Arturo

Aortic pathology

Small group discussion of subcommittees addressing specific research questions

12:00 – 13:00 – Lunch

13:00 – 15:30

Prioritization of MAC research projects

Other sites or investigators with specific expertise to invite to participate in MAC

Future MAC meetings

International Marfan Meeting - May 2018

Jo

16:00 – San Gimignano & Dinner

Friday, June 2, 2017

Breakfast / brunch and Departure