First 2 years of experience of an integrated multidisciplinary clinic for adults with aortopathies in a Canadian context

Anne-Marie Laberge[1-2], Ismail El Hamamsy[3], Laura Robb[1], Philippe Demers[3], Nancy Poirier[3], and François-Pierre Mongeon[1].

1) Département de médecine, Institut de Cardiologie de Montréal, Montréal, QC, Canada
2) Service de génétique médicale, Département de Pédiatrie, CHU Sainte-Justine, Montréal, Canada
3) Département de chirurgie cardiaque, Institut de Cardiologie de Montréal, Montréal, QC, Canada

BACKGROUND:

•Connective tissue disorders are multisystemic conditions affecting mainly the cardiovascular system, bones and joints, and the eyes.
• Diagnosis of Marfan syndrome requires a multisystemic evaluation, including a thorough physical examination and family history.
• Involvement of the heart and aorta can be life-threatening and therefore warrants specialized evaluation and management.

In 2012, the Montreal Heart Institute started an integrated multidisciplinary clinic for adults referred for suspicion of Marfan syndrome or other connective tissue disorders at risk of aortic disease, the Aortic and Connective Tissue Clinic (ACTC).
A heart team (cardiologist specialized in adult congenital heart disease, cardiac surgeons specialized in aortic surgery) and a genetics team (medical geneticist, genetic counselor) work side-by-side. Both teams see patients with a family history of aortic disease or systemic features of Marfan syndrome. The heart team sees patients with presumed isolated aortic disease and determines if evaluation by the genetics team is needed.

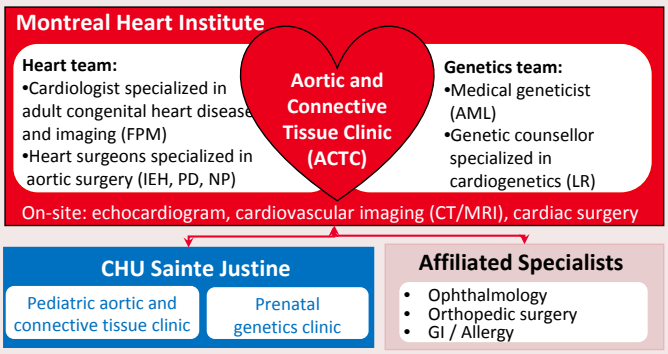

Figure. ACTC clinic structure and relationships with other specialized clinics

OBJECTIVE: Assess first two years of clinical activities

METHODOLOGY:

Data collection:
•Review of clinical database and electronic medical records and the cardiovascular genetics clinic's corresponding family files for all patients seen at the Aortic and Connective Tissue Clinic between May 2012 and June 2014 (n=165).
•Data was collected on referral context, cardiac evaluation, genetic evaluation including genetic tests, final diagnosis and planned follow-up.
Analysis: Descriptive statistics are reported

RESULTS: n= 165

Reason for referral (some had >1 reason for referral)	
Personal history of aortic dilation/dissection	53
Family history of aortic dilation/dissection	39
Clinical diagnosis of Marfan syndrome	31
Suspicion of Marfan syndrome	31
Family history of Marfan syndrome	12
Suspicion of Loeys-Dietz syndrome	4
Family history of Loeys-Dietz syndrome	2
Suspicion of other connective tissue disorder	16
Family history of sudden death	3
Other	21

Referred by (n=165)	
Physician within MHI	59
Physician from other center	58
Family member of patient seen in the clinic	11
Pediatric aortopathies and connective tissue clinic	12
Prenatal genetics clinic	4
Transfer to adult care by pediatric cardiology clinic	12
Self-referral/Other	9

63 seen by heart team only (5 already seen by genetics)

92 seen by heart and genetics teams

10 seen by genetics team only (4 already seen by cardiologist)

Average wait for 1st appointment: 3,8 months

Ongoing pregnancy when referred: 5 women and 1 male

14 adults referred after evaluation of affected child

10 children referred to pediatric clinic after evaluation of adult

Cardiovascular imaging	
Imaging:	138 patients had imaging at MHI 13 patients had imaging done only in other center 14 had no imaging
Echocardiogram	129 (+2 pending)
Cardiovascular MRI	51 (+7 pending)
CT aorta	11
Findings:	**N= 151**
Aortic dilatation (any level)	63 (42%)
Aortic dilatation at sinus of Valsalva	47 (31%)
Median and range	41 (37-65 mm)
Mitral valve prolapse	27 (18%)

Outcome of aortic disease	**Prior to 1st clinic visit**	**Since 1st clinic visit**
Acute events		
Acute aortic dissection	23	1
Other acute event	3 (2 coronary artery dissections, 1 abdominal aorta dissection)	0
Cardiovascular surgery		
Aortic surgery	28	8
Aortic surgery pending	--	1
Other cardiovascular surgery	6	1

Genetic evaluation	**n**	
Seen by medical geneticist	79	(including 11 seen by both)
Seen by genetic counselor	34	
Genetic test :		
Ordered at MHI	46	71 patients were tested overall, (4 were tested elsewhere and at MHI)
Done elsewhere prior to first visit	29	
Waiting for relative's result prior to testing	9	
Offered but declined	8	
No genetic test offered	77	
Genetic tests ordered at MHI: (some patients had > 1 test)		Results:
FBN1	33	9 pathogenic, 1 VUS (14 pending)
TGFBR1, TGFBR2, SMAD3, TGFB2	19	All normal (5 pending)
COL3A1	2	All normal
ACTA2	11	1 VUS likely pathogenic (3 pending)
Other	2	All normal (1 pending)

17 patients referred with known mutations: 8 FBN1, 3 TGFBR1, 1 TGFB2, 1 ACTA2, 1 EDS I, 1 MYLK, and 2 microdeletions

Final diagnosis	**N = 165**
Marfan syndrome (with FBN1 mutation)	17
Marfan syndrome (clinical diagnosis only)	15
Normal aorta with systemic score > 7	2
Loeys-Dietz syndrome	4
ACTA2 mutation	2
Familial TAAD	13
Ehlers Danlos syndrome (type I or type III)	5
Connective tissue disorder NOS	9
Aortic dilatation secondary to bicuspid aortic valve	7
Isolated aortic dilatation	17
No underlying connective tissue disease	24
Ongoing evaluation – no final diagnosis yet	47
Other	3

Planned follow-up	**n = 165**
Dismissed from clinic – no specific follow-up needed	23
Recommendations for follow-up given to referring physician	52
Follow-up at our integrated multidisciplinary clinic	90

CONCLUSIONS:

•Close links between our clinic and the pediatric ACTC as well as the prenatal genetic clinics have facilitated efficient cross-referrals .
- Links with affiliated specialists need to be strengthened.

•Our integrated multidisciplinary approach results in efficient access to specialized cardiac and genetic services:
- Short wait for first appointment
- 8 patients have had elective cardiovascular surgery since being seen
- 11 molecular diagnoses have been made, enabling familial screening

•26 patients had an acute arterial dissection prior to being referred:
- Increased awareness of Marfan syndrome and other connective tissue disorders could lead to earlier referral, better medical management and elective surgery when needed

Anne-Marie Laberge is supported by **CIHR**'s New Investigator Salary Award.