



Canadian Organization
for Rare Disorders

Amyloidosis
Program of Calgary



Unique Challenges of Diagnosis and Care for Adults with Rare Diseases

Canadian Organization for Rare Diseases Fall Conference

Nowell Fine, MD SM

November 29th 2023

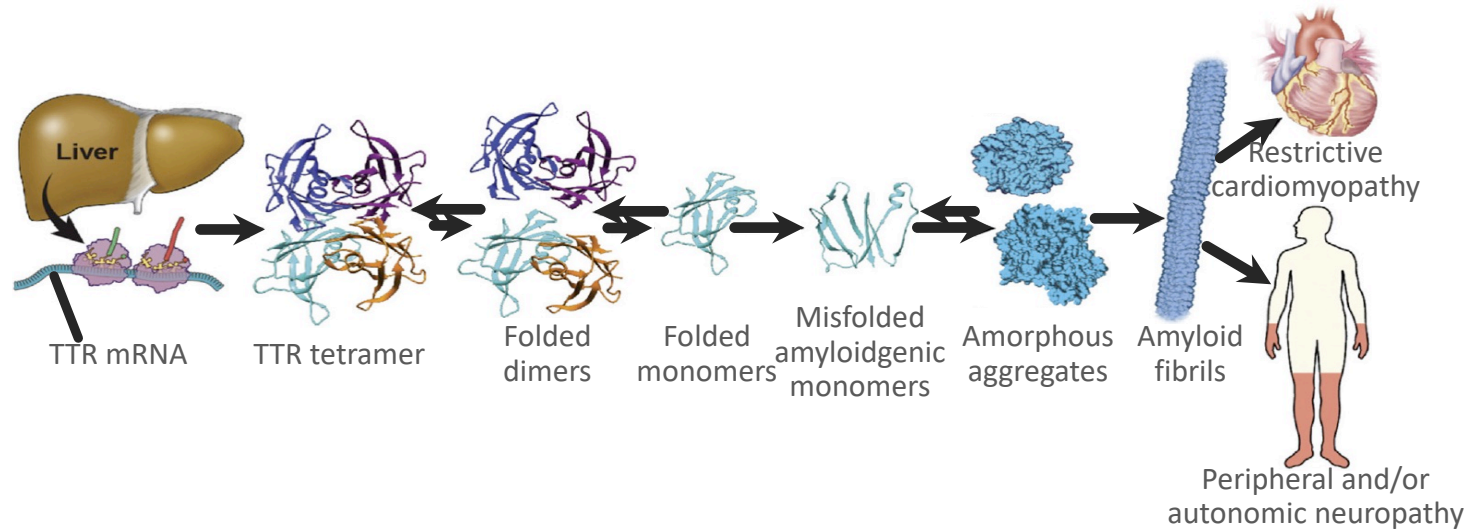


UNIVERSITY OF CALGARY
CUMMING SCHOOL OF MEDICINE

Disclosures

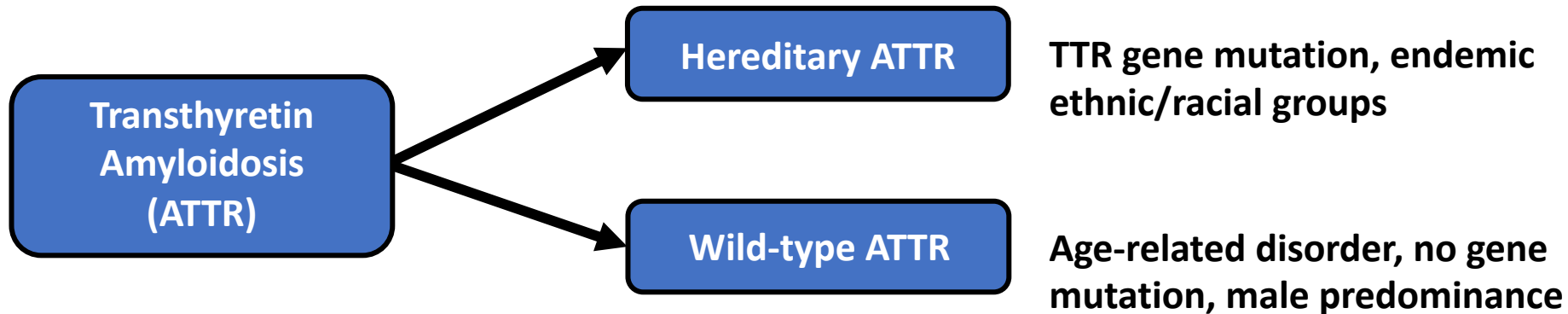
- **Grants/Research Support:** Pfizer, Ionis, Servier, Takeda, Novartis, BridgeBio-Eidos
- **Speaking/Consulting Honoraria:** Pfizer, Ionis, Sobi, Alnylam, Sanofi-Genzyme, Astra-Zeneca, Takeda, NovoNordisk

Transthyretin Amyloidosis (ATTR)



Diagnosed in older adults 70's, 80', 90's!

New (and expensive) therapies!



Diagnosis



Hiding in plain sight!

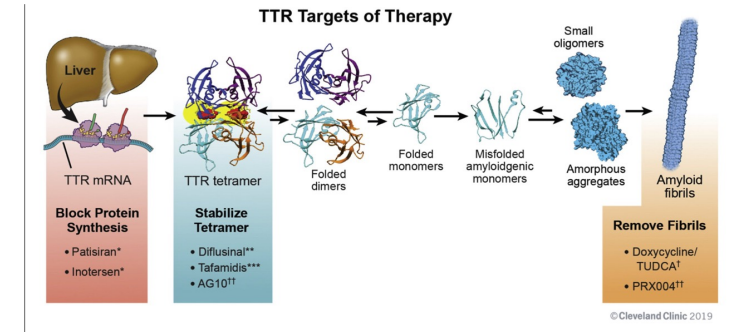
Clinical Care Issues



Comorbidities



Polypharmacy



Disease Modifying Therapy

- Projected benefit and lifespan

GOALS OF CARE DESIGNATIONS		Chest Compressions	Intubation	ICU Admit - Adult	ICU Admit - Pediatric	Surgery	Site Transfer	Symptom Control
R Resuscitative Care	1	✓	✓	✓	✓	✓	✓	✓
	2	X	✓	✓	✓	✓	✓	✓
	3	X	X	✓	✓	✓	✓	✓
M Medical Care	1	X	X	X	Can consider, if required for symptom control	✓	✓	✓
	2	X	X	X	Can consider, if required for symptom control	✓	✓	✓
C Comfort Care	1	X	X	X	Can consider, if required for symptom control	✓	✓	✓
	2	X	X	X	Can consider, if required for symptom control	X	X	✓

Goals of Care

Ageism



Multidisciplinary Care



It takes a village!



Avengers assemble!

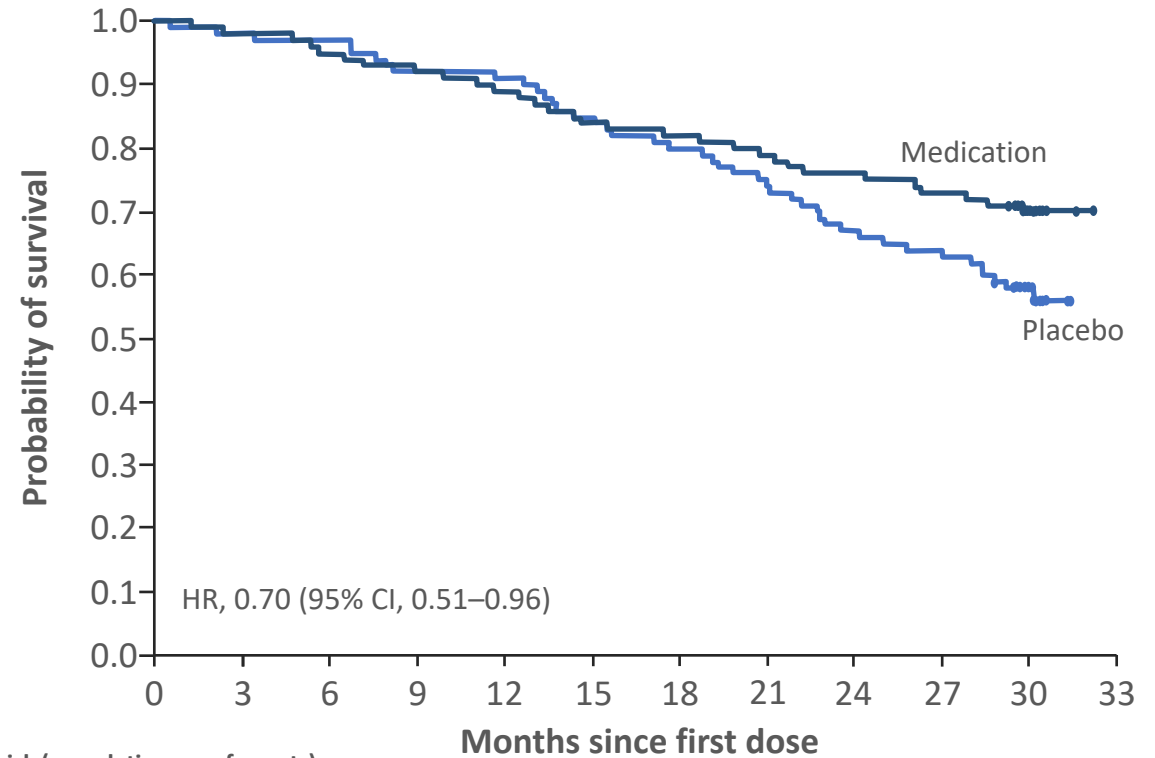
Genetic Testing



Drug Development



Mortality



No. at risk (cumulative no. of events)

	0	3	6	9	12	15	18	21	24	27	30	33
Medication	264(0)	259(5)	252(12)	244(20)	235(29)	222(42)	216(48)	209(55)	200(64)	193(71)	99(78)	0(78)
Placebo	177(0)	173(4)	171(6)	163(14)	161(16)	150(27)	141(36)	131(46)	118(59)	113(64)	51(75)	0(76)

Adult Rare Diseases

- Thank you!
- nmfine@ucalgary.ca



Canadian Organization
for Rare Disorders