

Supplementary Online Content

Pousset F, Legrand L, Monin ML, et al. A 22-year follow-up study of long-term cardiac outcome and predictors of survival in Friedreich ataxia. *JAMA Neurol*. Published online September 28, 2015. doi:10.1001/jamaneurol.2015.1855.

eTable 1. Characteristics of the 5 FRDA Patients With Heterozygous Point Mutations

eTable 2. FRDA Patients With Heterozygous Point Mutations: Follow-up and Survival

eTable 3. Cardiac Events and Outcome in 133 Friedreich Ataxia Patients During Follow-up

This supplementary material has been provided by the authors to give readers additional information about their work.

eTable 1: Characteristics of the 5 FRDA Patients With Heterozygous Point**Mutations**

	Patient 140-001	Patient 142-010	Patient 038-010	Patient 151-001	Patient 147-001
Point mutation	482+1G>C	317T>G	L182H	G130V	G130V
Shorter GAA repeats	500	767	800	833	967
Age (years)	19	35	14	44	43
Age at onset (years)	14	13	8	15	18
Age at wheelchair onset (years)		26	16	18	28
Left ventricular end diastolic diameter (mm)	45.0	46.0	36.5	42.0	46.0
Left ventricular end systolic diameter (mm)	28.0	27.0	18.4	24.0	30.0
Septal wall thickness (mm)	12.5	13.5	11.9	12.0	7.0
Posterior wall thickness (mm)	12.2	10.4	12.2	10.0	7.0
Indexed left ventricular mass (g/m ²)	111	117	103	82	67
Relative wall thickness	0.54	0.52	0.66	0.52	0.30
Left ventricular ejection fraction (%)	68	66	60	54	61
E/A ratio	1.02	1.73	1.81	0.55	1.51

eTable 2: FRDA Patients With Heterozygous Point Mutations: Follow-up and Survival

	Patient 140-001	Patient 142-010	Patient 038-010	Patient 151-001	Patient 147-001
Left ventricular ejection fraction <50%	no	no	yes (at age 16)	no	no
Last left ventricular ejection fraction during follow-up (%)	68	61	23	65	
Time between inclusion and last Left ventricular ejection fraction (years)	3.7	6.0	6.8	6.9	
Supraventricular arrhythmia	no	no	yes (at age 19)	no	no
Heart failure	no	no	yes (at age 18)	no	no
Death	no	no	no	no	no
Heart transplantation	no	no	yes (at age 21)	no	no

eTable 3: Cardiac Events and Outcome in 133 Friedreich Ataxia Patients During Follow-up

Events	FRDA Patients n (%)	Age (years)	Delay from disease onset (years)	Delay from events to death (years)
Supraventricular arrhythmia	22 (17%)	35 ± 10 [20-54]	24 ± 9 [12-38]	6.1 ± 3.1 [1.1-11.1] (n=11)
LVEF <50%	13 (10%)	34 ± 10 [22-60]	24 ± 9 [12-46]	5.3 ± 2.4 [1.1-8.8] (n=8)
Heart failure	11 (8%)	38 ± 12 [22-61]	27 ± 10 [14-47]	3.1 ± 2.3 [0-7.0] (n=7)
Death	15 (11%)	39 ± 10 [27-61]	30 ± 9 [17-45]	

Data are expressed as the mean ± standard deviation [min-max]