Disease characteristics and tissue frataxin concentrations in adults with Friedreich's ataxia participating in nomlabofusp interventional studies

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Methods: Disease characteristics (e.g. age at onset, guanine adenine adenine [GAA] repeat length) of adults with Friedreich's ataxia (FRDA) participating in Phase 1 and 2 nomlabofusp interventional studies were summarized and evaluated relative to baseline buccal and skin cell frataxin concentrations.

Results: Sixty-one subjects participated in at least one study; 18 participated in more than one study. Mean age was 31.9 years (range 19- 69). Mean age of onset was 15.9 years (range 5- 60). Mean (range) shorter and longer GAA repeat lengths were 555.8 (99- 1000) and 890.2 (265- 1300). Mean baseline modified Friedreich's ataxia Rating scale neurologic score was 49.5 (13.2- 74.5). Mean (range) baseline buccal and skin cell frataxin concentrations were 1.90 (0.70- 4.95) and 3.25 (1.40- 8.10) pcg/mcg, respectively. There is a relationship between frataxin concentrations and age of onset and GAA repeat length. There is also a relationship between skin and buccal cell frataxin concentrations.

Discussion: Early age of onset is associated with more rapid disease progression. Data from the nomlabofusp interventional studies are consistent with previously published data that suggest that lower tissue frataxin concentrations are associated with more rapid disease progression. Increasing frataxin concentrations in patients with FRDA may decrease the rate of disease progression.

Conclusion: The study population in the nomlabofusp interventional studies is representative of the FRDA population, and tissue frataxin concentration data from these studies are consistent with prior studies demonstrating that lower frataxin concentrations are associated with earlier onset of disease. Buccal and skin cell frataxin levels correlate with each other.