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Friedreich’s ataxia presenting after cardiac transplantation

A 4 year old boy underwent cardiac transplantation because of cardiomyopathy at the age of 6 months. He developed neurological symptoms associated with Friedreich’s ataxia. He underwent cardiac transplantation because of cardiomyopathy at the age of 6 months. He developed neurological symptoms associated with Friedreich’s ataxia. The incidence of Friedreich’s ataxia is in 1/50,000 and typical presenta-
tion is before 25, with gait or stance ataxia and dysarthria, lower limb areflexia, and an extensor plantar response. Reduced vibration sense, pes cavus, and scoliosis are often present.5 6

Cardiomyopathy is frequent later in the course of the disease.7 Triplet repeat lengths range from 7 to 29 in normal people and 66 to 1360 in Friedreich’s ataxia patients.8

Since the location of the frataxin gene in 19889 and availability of genetic testing for Friedreich’s ataxia, a broader clinical spectrum in terms of age and presenting features is now recognised.9 10 Early onset and rate of progression are predicted by large expansions.11 However, it is very unusual for cardiomyopathy to be the presenting feature and we can identify only five cases described in the literature where this has occurred.12 13 None were as young as our patient, in keeping with the large repeat length in this child.

To our knowledge this is the first case published of a patient with Friedreich’s ataxia undergoing cardiac transplantation. This is probably because cardiomyopathy usually occurs after neurological disease is well established and the poor prognosis precludes transplantation. Our patient is unusual in that he had early life threatening cardiomyopathy but mild neurological disease. If we had recognised the genetic abnormality before transplantation, the prognosis of neurological disease and survival after cardiac transplantation (50–60% at 10 years)13 would have been discussed with his parents before making a decision with them. The cardiac transplantation has almost certainly improved his life expectancy.