Hypertrophic cardiomyopathy in combination with juvenile amaurotic idiocy

Chance or fundamentally related findings?

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SUMMARY Hypertrophic cardiomyopathy and juvenile amaurotic idiocy (one of the ceroid lipofuscinoses) were diagnosed in a 29 year old man. This combined finding may be one of pure coincidence, but hypertrophic cardiomyopathy like changes of the myocardium are known to occur in Friedreich's ataxia and lentiginosis. The occurrence may, therefore, indicate some fundamental interrelation.

Curiosity is always aroused when two rare disorders are present in one patient. Is it pure coincidence or does it signify some fundamental connection? Hypertrophic cardiomyopathy is an uncommon form of familial myocardial disease with an autosomal dominant trait of inheritance. Its incidence is unknown. Juvenile amaurotic idiocy belongs to the so called ceroid lipofuscinoses and is of autosomal recessive inheritance. The incidence is said to be low, perhaps 1-3 per 100 000 newborn per year.¹ We report a case in which both disorders were present.

Case report

Since the age of 4-5 years, a 29 year old man had shown the classical signs of juvenile amaurotic idiocy, which are progressive visual failure leading to blindness, extrapyramidal signs and seizures, and increasing mental retardation. Vacuolated lymphocytes were present in the peripheral blood. At the age of 13 years a faint systolic murmur was heard at the left sternal border, and at 20 years the electrocardiogram showed left ventricular hypertrophy (Figure). There were no objective signs of cardiac failure. At 29 years the patient died of bronchopneumonia and respiratory failure.

At necropsy the heart weighed 415 g (the patient's

height had been 175 cm and weight 45 kg). The heart appeared to be of normal form with no congenital abnormalities and without external calcification. The septum was not asymmetrically hypertrophied. Microscopical examination showed large amounts of lipopigment in the myocardial cells but neither cholesterolic compounds nor dystrophic calcification as seen in most cases of juvenile amaurotic idiocy.¹ Fibre disarray was present in about 40% of the left ventricle. This was estimated by determining the volume fraction of disarrayed fibres (point counting) in a series of sections including the entire circumference of the left ventricular cavity at mid-ventricular level. The alterations were dispersed concentrically and also involved the posteromedial part of the right ventricle. The histological hypertrophic cardiomyopathy index² totalled 66% with a pronounced degree of cellular fibrosis and fibre disarray (Figure) (fibrosis, 3 points, bizarre nuclei, 2, perinuclear halos, 0, whorls, 2, and short fibres, 3, a total of 10 points (66%) (0 point, 0%; 15 points, 100%)).

The brain was atrophic and weighed 950 g. The cerebral cortex was thin and of a peculiar yellowish brown colour. There was a general loss of ganglionic cells, which contained a great amount of lipopigment. There were no calcifications in the cerebral tissue; slight calcification of the meninges and vessels was evident. The striated muscles showed neurogenic atrophy, and only very little lipopigment was present in these cells. Thus evident features of juvenile amaurotic idiocy and hypertrophic cardiomyopathy were present.

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Figure (a) Electrocardiogram showing inverted T waves in V4 and V6. (b) Histological section of left ventricular myocardium with disorganised muscle fibres, short runs, hypertrophy, and fibrosis (haematoxylin and eosin $\times 100$). The insert shows black areas, which indicate where fibre disarray was found in a transsection of the heart (mid-ventricular).

Discussion

This patient reached the age of 29 years. This is unusual, since most patients with juvenile amaurotic idiocy die 6–10 years after the time of diagnosis. The probable reason is that he was very well cared for by his family.

We have already reported the structural findings of the heart in a series of 13 patients with juvenile amaurotic idiocy.¹ In general, the heart was enlarged, and various electrocardiographic changes were present in 10. In none of these patients were features of hypertrophic cardiomyopathy present. Olsen has repeatedly stated that fibre disarray by itself is no determinant of hypertrophic cardiomyopathy; only when a certain pattern and extent of structural changes are present—as expressed by the so called histological index²—can hypertrophic cardio-

myopathy be firmly diagnosed. In cases without obstruction the fibres are usually distributed throughout the left ventricular or right ventricular wall or both, as was the case in the present patient. The concentric form of hypertrophic cardiomyopathy is rare but well recognised; ample evidence shows that morphological criteria for obstructive and non-obstructive cases are not altogether reliable.² In the present patient all criteria for a diagnosis of both juvenile amaurotic idiocy and concentric hypertrophic cardiomyopathy were fulfilled.

The association between Friedreich's ataxia and hypertrophic cardiomyopathy like changes is well documented,² as is a relation between lentiginosis and hypertrophic cardiomyopathy.³ What Friedreich's ataxia, lentiginosis, and juvenile amaurotic idiocy have in common is not at all clear. They may all be metabolic disorders or due to dysfunction of embryonic neural tube/neural crest elements or both.

The occurrence of two rare disorders in one patient has often led to the assumption that such disorders are linked in a fundamental way. Because of the low prevalence of two rare entities, their presence in the same patient would be an unexpected chance finding. It would, however, be unreasonable to draw such a conclusion on the basis of a single case report, but we believe it is justified to add our findings to those already collected on hypertrophic cardiomyopathy. These observations may prove the two conditions are related in some way (hypertrophic cardiomyopathy may occasionally be the end result of several different metabolic abnormalities and therefore an unusual manifestation in protracted cases of juvenile amaurotic idiocy) or to be two unrelated entities.

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References

- 1 Reske-Nielsen E, Baandrup U, Bjerregaard P, Bruun I. Cardiac involvement in juvenile amaurotic idiocy—a specific heart muscle disorder. Histological findings in 13 autopsied patients. *Acta Pathol Microbiol Scand [A]* 1981; 89: 357–65.
- 2 Olsen EGJ. *The pathology of the heart*. 2nd ed. London: Macmillan, 1980: 317–50.
- 3 Somerville J, Bonham-Carter RE. The heart in lentiginosis. *Br Heart J* 1972; 34: 58–66.