Genetic miscounselling in muscular dystrophy

Sir,

Raised serum creatine phosphokinase (CPK) is now widely used in the diagnosis of Duchenne muscular dystrophy and in detection of female carriers. While a significant rise in CPK in the potential carrier is presumptive confirmation of her carrier status, a normal result does not exclude it. About 20–30% of genetically definite carriers have consistently normal enzyme levels. In addition, the level of CPK may fluctuate in the same individual and it is advisable to repeat the examination on at least three occasions. In possible carriers with a normal CPK level additional help in confirmation of carrier status may be obtained from overt abnormality on quantitative electromyography or muscle biopsy (for review see Dubowitz, 1975).

Case report

A 19-year-old female with 2 brothers who had died of classical Duchenne muscular dystrophy at the ages of 22 and 20 years, respectively, sought genetic counselling at her local hospital at the time of her marriage. On the basis of a single normal serum CPK result she was advised that she was not a carrier and could safely have children. After a period of prolonged infertility she became pregnant 7 years later after clomiphene stimulation and artificial insemination and delivered a normal male infant at term.

A cord blood sample for CPK was not obtained but a venous sample from the infant on the 3rd day showed a level >1000 IU/l; at the age of 6 weeks two samples were 5000 and 4900 IU/l, respectively; and at 7 weeks >1000 IU/l. The mother’s levels at 4 and 5 weeks post partum were 95 and 60 IU/l. (Method used: Boehringer kit; normal for adults 0–130 IU/l) At the age of 11 weeks the infant’s CPK was 1090 IU/l, and that of the mother 22 IU/l. (Method: Boehringer (25°C), normal for adult female <60 IU/l.) Electromyography on the infant was normal but a muscle biopsy from the rectus femoris showed unequivocal changes of early dystrophy.

This case illustrates the difficulty of excluding Duchenne dystrophy in a possible female carrier with normal CPK levels. As she already had 2 affected brothers one can assume that her mother was a definite carrier and she thus already had potentially a 50% chance of being a carrier on genetic grounds. In such instances it is advisable for patients to be referred to special centres for more detailed assessment of their carrier status.

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REFERENCE


Insulin response to glucagon in short children

Sir,

Dr. Karp and his colleagues have reported low insulin responses to glucose and arginine stimulation in thin children with familial short stature and have recently shown that glucagon injection produces a normal rise in insulin in these patients (Karp, Laron, and Doron, 1975). It is possible that the poor insulin response to glucose in these patients may be secondary to their growth failure, rather than an aetiological factor. There is evidence that serum insulin levels after oral glucose vary with age and low levels are common in small children (Grant, 1968; Paulsen, 1973; Rosenbloom et al., 1975). The explanation for this is not known but it is possible that sensitivity to endogenous insulin falls with increasing maturity. The low insulin responses to glucose which were found by Karp et al. in their patients may be related to physical ‘immaturity’, and it would be most interesting to learn whether the results were still abnormal when compared with those obtained in normal children of comparable size and skeletal maturity.

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REFERENCES


