Correspondence

Journal of Medical Genetics 1989, 26, 216

Absent or hypoplastic extraocular muscles?

Sir,

I read with fascination the report ‘Absence of a lateral rectus muscle associated with duplication of the chromosome segment 7q32→q34’ (Keith CG, Webb GC, Rogers JG. J Med Genet 1988;25:122–7).

Recent reports1-3 of extraocular muscle deficiencies of this nature have shown that there is almost always an extraocular muscle in the orbit, perhaps rudimentary or small, but that true absence may well be an extraordinarily rare event, if it ever does, in fact, occur in an absolute manner—especially in the presence of a relatively normal eye and orbital content as in this case.

Would the authors of this article advise us as to whether or not computerised tomography was carried out on this patient, and if so, was there evidence of a lateral rectus muscle in the orbit on the right side? If no such study was done, I would recommend that the authors obtain such a study, to define more accurately the abnormalities of this syndrome. A CT scan might even be useful in the management of the residual strabismus which the patient has. In the near future, in such cases, strabismus surgeons will be going back into the orbit and attaching prosthetic tendons from the globe to such rudimentary muscles. Such might even be attempted in the case reported, if an orbital muscle of reasonable dimension and location is found on CT scan.

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References


This letter was shown to Mr C G Keith, who replies as follows:

Sir,

We are grateful for Dr Romano’s interest in our paper and for calling our attention to the usefulness of CT scanning for muscle detection in the orbit. The child did have a CT scan of the brain and orbits, but unfortunately the slice level was set at 37.5 mm, so only one slice was obtained through the orbit and it was not possible to determine from this whether the muscle was in fact present. In view of the child’s general condition it was not considered reasonable to repeat the CT scan and, since then, the child has died. CT scanning has been used in our department routinely in checking for slipped or lost muscles and it was used to show the absence of the lacrimal gland in a case of alacrimia.1 Dr Romano raises a very important point and perhaps we should rename the paper ‘Congenital non-attachment of the lateral rectus’.

C G Keith
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Reference

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