Hypoplastic left heart in cerebrocostomandibular syndrome

Cerebrocostomandibular syndrome (CCMS) is a rare disorder characterised by severe micrognathia and posterior "rib gap" defects. Since the first report of this condition by Smith et al. in 1966, 50 cases have been reported. 1 Severe micrognathia and radiographic evidence of posterior rib gap defects have been constant features. We report a female infant with typical features of CCMS who also had hypoplastic left heart syndrome, which caused her death. A cardiac lesion has been described in only one family, and an infant with a large ventricular septal defect. 1

The female proband was the third child of healthy, unrelated parents. Her father and mother were of French and Mauritian descent, respectively. Neither parent had any clinical or radiographic evidence of mild cardiac malformations even though this finding is uncommon.

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Another dystonia

I read the Syndrome of the Month article on the dystonias by Jarman and Walker with great interest. 1 This is a difficult clinical subject to study and the move to a molecular genetic classification is to be applauded. Accordingly I would like to draw attention to another form of dystonia which has been mapped to the short arm of chromosome 11.

The dystonia is manifest mainly in the hands. The onset can be recognised in childhood by an odd positioning for some voluntary movements such as holding a pencil; this has been called "fisting" by the family. More obvious dystonic movements develop in adolescence and these progress slowly over the years. The hands of the oldest affected subject, now aged 68, are severely disabled and he needs to be dressed and fed. This man also has dysarthria.

The other prominent and important feature of this syndrome is mental retardation of mild to moderate degree, more often the latter. The heterozygote carriers do not seem to be affected.

The syndrome is quite well known in medical genetics publications. 1 It has been given two gene symbols, PRTS' and MRXS1, and is listed in the McKusick catalog (MIM 309510). It was first reported 10 years ago in one family but so far no other cases have come to light. It is conceivable that it is a private syndrome with the mutation occurring in this family alone, but I believe it more likely that there are others with the same disorder which have just not been recognised. Part of the reason may be that the neurologists are not familiar with this form of dystonia.

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