were healthy, clinical descent, respectively. have been female infant with as a viewed some The infant with an Cerebrocostomandibular syndrome C282Y point relatively unexpected high frequency a viewed against some 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.

Another dystonia

I read the Syndrome of the Month article on the dystonias by Jarman and Walker with great interest. 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 MRXS1,4 and to the short arm of chromosome X chromosomes between DXS5 and DXS553.1

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. It has been given two gene symbols, PRTS1 and MRXS1, and is listed in the McKusick catalogue (MIM 309510). It was first reported 10 years ago in one family in Algeria, 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.

M W PARTINGTON
Hunter Genetics, PO Box 84, Warratah, NSW 2298, Australia


Figure 1 Chest radiograph showing multiple rib gaps.

CCMS. The pregnancy was uncomplicated, with spontaneous vertex delivery at 37 weeks' gestation. Birth weight was 2200 g, length 45 cm, and occipitofrontal circumference 31 cm (all less than the 3rd centile for gestational age). Features noted at birth included severe microretrognathia, glossoptosis, and a midline cleft of the soft palate, all of which contributed to early upper airway obstruction and moderate respiratory distress. Initial examination was otherwise normal.

A chest radiograph (fig 1) showed multiple posterior rib gaps involving the second to sixth ribs on the right and the second to fifth ribs on the left. The gap in the right second rib resembled a pseudarthrosis. There were 11 pairs of ribs. Cranial ultrasound was normal. Karyotype on peripheral blood lymphocyte culture was 46,XX.

On the third day of life a cardiac murmur was noted. Echocardiography showed findings consistent with hypoplastic left heart syndrome. Given the poor prognosis associated with hypoplastic left heart syndrome, active management was withdrawn and she died aged 8 days. Her parents declined necropsy.

This is the second reported case of a cardiac anomaly in CCMS, and the first in which hypoplastic left heart syndrome is described. A potential mechanism to account for the concurrence of hypoplastic left heart and CCMS is unclear. Vascular insufficiency has been postulated in the causation of hypoplastic left heart syndrome, but there is no evidence to support a disturbance in normal rib morphology on this basis. Embryologically, the heart is derived from lateral plate mesoderm and the ribs from paraxial mesoderm. Mandibular deficiency is considered to be the result of a defect in the ventral portion of the first branchial arch secondary to defective neural crest cell migration or proliferation. CCMS and hypoplastic left heart syndrome in our patient may have occurred together as part of a developmental field defect. Alternatively, an underlying basic defect in a dermato-epidermal junction control or signalling gene is possible. The CCMS phenotype should be expanded to include cardiac malformations even though this finding is uncommon.

EDWIN KIRK
LESLEY ADES
Departments of Clinical Genetics, and Paediatrics and Child Health, Royal Alexandra Hospital for Children, Hawthorne Road, Westmead, NSW 2145, Australia


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 identified in only one previous CCMS infant, 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
Hypoplastic left heart in cerebrocostomandibular syndrome.

E Kirk and L Ades

*J Med Genet* 1998 35: 879
doi: 10.1136/jmg.35.10.879

Updated information and services can be found at:
[http://jmg.bmj.com/content/35/10/879.1.citation](http://jmg.bmj.com/content/35/10/879.1.citation)

These include:

**Email alerting service**
Receive free email alerts when new articles cite this article. Sign up in the box at the top right corner of the online article.

Notes

To request permissions go to:
[http://group.bmj.com/group/rights-licensing/permissions](http://group.bmj.com/group/rights-licensing/permissions)

To order reprints go to:
[http://journals.bmj.com/cgi/reprintform](http://journals.bmj.com/cgi/reprintform)

To subscribe to BMJ go to:
[http://group.bmj.com/subscribe/](http://group.bmj.com/subscribe/)