Kenny-Caffey syndrome without the CATCH 22 deletion

With regard to the recent report by Sabry et al, we would like to present an additional case of Kenny-Caffey syndrome who, unlike their case, did not have a chromosomal deletion at 22q11.2 (the CATCH 22 region). We also report detailed test results for calcium metabolism and response to combination therapy with vitamin D, magnesium, and growth hormone.

The female patient was born after 40 weeks of an uneventful pregnancy to non-consanguineous, healthy, Japanese parents. She weighed 2750 g and measured 46 cm at birth. At 1 month, she had an episode of generalised convulsions because of hypocalcaemia. During this episode, her serum calcium, phosphorus, magnesium, calcium x phosphorus product and intact PTH were 5.0 mg/dl (reference range 8.6-9.7), 9.1 mg/dl (2.7-4.3), 1.2 mg/dl (1.8-2.2), 120 pg/ml (94-156), and <15 pg/ml (15-50), respectively. Oral 1α-hydroxyvitamin D (0.3 μg/day) was started on the basis of a diagnosis of hypoparathyroidism.

The patient’s serum calcium levels were normalised although her intact PTH remained at low to low-normal levels. She had another episode of hypocalcaemic convulsions at 9 months of age. At the age of 5 years 1 month, she was referred to our hospital for further examination.

Physical examination showed her to be of proportionally short stature. Her height was 84.2 cm (mean ±5.3 SD) and her weight was 12.2 kg. She had normal intelligence, a prominent forehead, and slender extremities. Her anterior fontanelle still showed an opening of 1 x 1 cm. She had severe hyperopia and normal optic fundi. Radiological examination showed medullary stenosis of the long bones typical of Kenny-Caffey syndrome. CT scan of the brain showed fine calcification in the basal ganglia. Although her serum calcium remained normal with relatively low doses of vitamin D, the EDTA loading test (50 mg/kg) indicated a blunted response for i-PTH (basal 12.8 pg/ml, peak 17.5 pg/ml). The PTH loading test showed a normal response to exogenous PTH with increased urinary excretion of phosphorus and cAMP. Growth hormone provocative tests showed normal responses.

As in the case of Sabry et al, we also suspected the possibility of the patient having a deletion in the CATCH 22 region. However, FISH analysis on peripheral blood lymphocytes (probe D22S29, and in situ hybridisation) showed a normal diploid state. The patient was then put on a combination therapy of vitamin D (1 μg/day) and magnesium sulphate (0.4 g/day). Since her short stature did not improve with this therapy, growth hormone therapy (0.5 IU/kg/week) was initiated at the age of 7 years 5 months. After two years of this therapy, the height standard deviation for her chronological age improved from −5.4 SD to −4.4 SD without appreciable acceleration of bone maturation.

The reason for the discrepancy between the Bedouin family reported by Sabry et al and our case remains unclear. One possibility is the genetic heterogeneity of the syndrome.

Kenny-Caffey syndrome is a rare autosomal recessive disorder characterized by short stature, mental retardation, and skeletal abnormalities. Despite the rarity of the disorder, several reports have described patients with similar clinical features. In this case, the patient had a diagnosis of Kenny-Caffey syndrome based on clinical and laboratory findings. The patient responded well to combination therapy, including vitamin D, magnesium, and growth hormone, which improved her calcium levels and growth parameters.

The patient's case highlights the importance of careful monitoring and management of calcium metabolism in patients with Kenny-Caffey syndrome. Further studies are needed to understand the underlying mechanisms and to improve the treatment strategies for this condition.

JUDITH GOODSHIP
Division of Human Genetics, University of Newcastle upon Tyne, 19/20 Claremont Place, Newcastle upon Tyne NE2 4AA, UK

Kenny-Caffey syndrome is part of the CATCH 22 haploinsufficiency cluster.

J Goodship

*J Med Genet* 1998 35: 1054
doi: 10.1136/jmg.35.12.1054-a

Updated information and services can be found at:
[http://jmg.bmj.com/content/35/12/1054.2.citation](http://jmg.bmj.com/content/35/12/1054.2.citation)

**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/)