A case of Kenny-Caffey syndrome

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We report a case of a patient with Kenny-Caffey syndrome who presented with hypocalcaemic seizures during his neonatal period due to hypoparathyroidism. The clinical features of this syndrome consist of growth retardation, medullary stenosis of the long bones, intermittent or long-standing hypocalcaemia due to mild to complete hypoparathyroidism, eye abnormalities such as microphthalmia, hyperopia, myopia and normal intelligence. Some of the clinical manifestations may be similar to pseudohypoparathyroidism. To recognize this syndrome early, we suggest a radiological study of the long bones in neonates with hypocalcaemia and wide anterior fontanel.

Key words: Kenny-Caffey, Hypoparathyroidism.

Reprint request: Wacharasindhu S, Department of Paediatrics, Faculty of Medicine, Chulalongkorn University, Bangkok 10330, Thailand.

Received for publication. April 9, 1998.

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กลุ่มอาการ Kenny - Caffeey : รายงานผู้ป่วย 1 ราย. จุฬาลงกรณ์มหาวิทยา 2541 มิ.ย. 42(6) : 455-60

รายงานผู้ป่วย Kenny - Caffeey syndrome 1 ราย ที่มีพัสดุอาการซึ่งในวัยแรกคลอด ขนาดจาก
ภาวะแคลเซียมในเลือดสูง ที่มีสาเหตุจาก hypoparathyroidism จากการขาดการแทรกซึมของโรคกลุ่มนี้
ได้แก่ growth retardation, delayed closure of anterior fontanel, medullary stenosis ของ long bone,
hypoparathyroidism ความมีโรคปกติทางตา ได้แก่ microphthalmia, hyperopia, myopia ระดับเด็กปฐมวัย,
ในเด็กกลุ่มนี้มีภาวะปกติ ดังนั้นในเด็กแรกเกิดมีภาวะแคลเซียมในเลือดสูงในเด็กที่ร่วมกับกัมภาพของ anterior
fontanel วั้ง ควรตรวจทางรังสีของกระดูก long bone เพื่อให้ได้การวินิจฉัยกลุ่มอาการได้เร็วขึ้น
Kenny-Caffey syndrome is an extremely rare disorder characterized by hypocalcemia, internal cortical thickening and medullary stenosis of tubular bones, delayed closure of anterior fontanel, eye abnormalities, short stature and normal intelligence. This condition was first reported in 1966 by Kenny and since then there have been 27 cases reported in the literature. Although most cases of Kenny-Caffey syndrome are thought to be inherited as an autosomal dominant, some cases may be autosomal recessive or sporadic. We report a case of this disorder presented during the neonatal period with hypocalcemic seizures due to hypoparathyroidism.

Case report

An 8-day old Thai boy was admitted to our hospital because of many episodes of jerking movement of facial muscles followed by the same in the upper and lower extremities, more on the left side, without conscious changes. Each attack lasted about 2 minutes. He was the first offspring of nonconsanguineous parents and was delivered by cesarian section due to placenta previa. His birth weight was 3200 gm. There were no remarkable events during the first week of his life. There was no history of this condition in his parents.

The initial physical examination upon admittance revealed a weight of 3300 gm and length of 51 cm. He had a prominent forehead with the anterior fontanel measuring 7 x 8 cm, the posterior fontanel measuring 3 x 3 cm and a head circumference of 34 cm. No other abnormalities were detected from physical examination.

The results of the laboratory investigations were as follows. The complete blood count revealed, hematocrit 48.7%, hemoglobin 15.6 g/dl, white blood cell 18,340 mm^3, PMN 38%, L 45%, M 10%, E 4%, B 3%, platelets 433,000/mm^3. Normal urine findings were found. Serum glucose 4.4 mmol/L, Na 136, K 5.7, Cl 105, HCO3 19 mmol/L, ionized calcium 0.9 mmol/L, phosphorus 3.7 mmol/L, magnesium 0.6 mmol/L, albumin 42 g/L, urea nitrogen 1.4 mmol/L, and creatinine 35.3 μmol/L. The cerebrospinal fluid from a lumbar puncture was normal.

The radiological study of the long bones showed cortical thickening and medullary stenosis and absence of a diploic space was demonstrated on skull X-ray (Figure 1,2) . Because of seizures with hypocalcemia, he was initially treated intravenously with 10% calcium gluconate. Subsequently, he was put on elemental calcium supplement orally up to a dose of 100 mg/k/day (Calcium-Sandoz - 1 tablet contains 2.94 g of Ca lactate-gluconate, 0.3 g of Ca carbonate equivalent to 500 mg of elemental calcium) and calcitriol (Rocaltrol) 100 ng/kg/day because of suspicion of hypoparathyroidism. With this treatment, the serum calcium and phosphate levels were normalized and the patient was free of seizures. Finally, the diagnosis of hypoparathyroidism was confirmed by a low serum intact parathyroid hormone level of 0.1 pmol/L (IRMA; normal range 1.4-5.7).

Discussion

The clinical constellation of 1) medullary stenosis of the long bones 2) growth retardation, 3) intermittent to long-standing hypocalcaemia due to mild to complete hypoparathyroidism, and 4) ophthalmologic abnormalities should make anyone suspicious of Kenny-Caffey syndrome. However,
Figure 1. The radiological study of long bone demonstrates cortical thickening and medullary stenosis.

Figure 2. Skull X-ray demonstrates the absent of diploic space in this patient.

diagnosis in the neonatal period may be difficult because some clinical manifestations such as growth retardation and eye abnormalities may not be presented during this period. In addition, X-rays of the long bones are not routinely performed for hypocalcemic seizures. The condition may be misdiagnosed as pseudohypoparathyroidism because of the short stature, and hypocalcemia suggests a spectrum of pseudohypoparathyroidism. However, the shortened fourth metacarpal bone and metastatic subcutaneous calcification would not. Franceschini et al (2) reviewed the previously reported cases and showed that more than 90% of the patients had short statures, cortical thickening with medullary stenosis of the long bones and delayed fontanelle closure, but three patients reported by Majewski et al (6) showed no medullary stenosis. Eighty five percent of reported cases had hypocalcemia, but low parathyroid hormone was found in only 60%. Boynton et al (7) also reported a Kenny-Caffey patient with no parathyroid tissue at autopsy at 19 years of age. Other abnormalities can be found in this disorder, such as microphthalmia (56%), hyperopia (61%), myopia (21%), and delayed dentition (67%). Eventually, most of the patients became of short stature with normal growth hormones (GH) and thyroid hormones. Short term GH therapy proved to be ineffective in this condition. Up to the present, no evidence of pituitary causes of the short stature have been report. studies in women with this condition showed normal sexual maturity and normal fertility, however, cryptorchidism was reported in a man with Kenny syndrome. More than half of the cases are familial, probably transmitted as autosomal dominants but autosomal recessive and sporadic cases have also been reported in molecular study. Bergada et al demonstrated no parathyroid hormone gene abnormalities in this condition. Intelligence was reported to be normal in 90% of cases in spite of many episodes
of seizure.\(^{(4)}\) Furthermore, iron deficiency anemia could be demonstrated in this condition but anemia related to medullary stenosis has never been proven.\(^{(2)}\) Table 1 shows the relative incidence of major manifestations in previously published cases of Kenny-Caffey syndrome compared with those in our patient. Because of a high percentage of delayed closures of fontanel, we suggest that X-ray of the long bones should be performed in hypocalcaemic neonates with wide anterior fontanel so as to recognize this condition early.

**Table 1. Relative incidence of major manifestations of Kenny - Caffey syndrome.**

<table>
<thead>
<tr>
<th>Major manifestations</th>
<th>Previous published cases</th>
<th>Our patient</th>
<th>Incident at present (%)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Sex, M:F</td>
<td>12:15</td>
<td>M</td>
<td>13:15</td>
</tr>
<tr>
<td>Cortical thickening and medullary stenosis</td>
<td>26/27</td>
<td>present</td>
<td>27/28 (96 %)</td>
</tr>
<tr>
<td>Dwarfism</td>
<td>25/27</td>
<td>not present</td>
<td>25/28 (89 %)</td>
</tr>
<tr>
<td>(need long term follow up)</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Delayed fontanelle closure</td>
<td>21/23</td>
<td>present</td>
<td>22/24 (92 %)</td>
</tr>
<tr>
<td>Hypocalcemia</td>
<td>19/22</td>
<td>present</td>
<td>20/23 (87 %)</td>
</tr>
<tr>
<td>Eye abnormalities</td>
<td>17/25</td>
<td>not present</td>
<td>17/26 (65 %)</td>
</tr>
<tr>
<td>Normal intelligence</td>
<td>18/20</td>
<td>need long term follow up</td>
<td>18/20 (90 %)</td>
</tr>
<tr>
<td>Low serum parathyroid hormone</td>
<td>7/12</td>
<td>present</td>
<td>8/13 (61 %)</td>
</tr>
</tbody>
</table>

**References**


9. Wilson MG, Maroni RF, Mikity VG, Shinno NW. Dwarfism and congenital medullary stenosis (Kenny syndrome). Birth Defects 1974;10(12):129-32