Kenny-Caffey syndrome is a rare hereditary skeletal syndrome characterized by dysmorphic features, severe growth retardation, classical radiological changes and hypocalcemia with hypoparathyroidism at an early age. We report an 8-month-old girl child with Kenny-Caffey syndrome who had most of the features of the syndrome. Any child with hypocalcemia who has typical facial features should raise a suspicion of this syndrome.

**Key words:** Growth retardation, hypoparathyroidism, Kenny-Caffey syndrome

**Introduction**

Kenny-Caffey syndrome is a rare syndrome characterized by dysmorphic features, growth retardation, uniformly small slender long bones with thickened cortex and medullary stenosis. Hypocalcemia with hypoparathyroidism presents at an early age. We report an 8-month-old girl child with Kenny-Caffey syndrome who had all the characteristic clinical, biochemical and radiological abnormalities of the syndrome.

**Case Report**

An 8-month-old female infant presented with seizures since 7 days of age. She was the third child born to consanguineous parents at term by cesarean section in view of fetal distress and breech presentation in a private hospital. Her birth weight was 2.3 kg and she cried at birth. On the seventh day of her life, she developed multifocal seizures and was detected to have hypocalcemia. Serum PTH done at that time was very low. She has been on calcium and vitamin D supplements since then. She was thriving poorly and she had not yet attained head control when first seen by us.

On examination, her weight was 5 kg, length was 62 cm and head circumference was 42 cm. She had a wide open anterior fontanel (5 × 5 cm), frontal prominence, depressed nasal bridge, long philtrum, micrognathia, up-slanting and deep-set eyes. Her ear lobules were underdeveloped [Figure 1]. Systemic examination including the cardiovascular system was within normal limits. Ophthalmic examination did not reveal any abnormality.

Investigations revealed hemoglobin 7 gm%, serum calcium 8.1 mg%, serum phosphorus 9.5 mg%, serum magnesium 1.61 mg% and serum alkaline phosphatase 355 U/L. Renal function was normal. Serum PTH was sent in view of hypocalcemia and hyperphosphatemia persisting in spite of supplementation. Her plasma parathyroid hormone level was reported as 3.68 pg/ml (normal 10-70 pg/ml), confirming our suspicion and

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**Figure 1:** Girl baby with Kenny-Caffey syndrome showing frontal prominence, depressed nasal bridge, long philtrum, micrognathia, up-slanting and deep-set eyes; her ear lobules were underdeveloped
suggesting a more severe underlying developmental abnormality. Neonatal hypoparathyroidism is usually transient; permanent congenital hypoparathyroidism is very rare. Most cases are caused by defective embryogenesis of structures arising from third and fourth pharyngeal pouches and fourth branchial arch. The commonest clinical syndrome known to occur is the third-fourth pharyngeal pouch syndrome or DiGeorge syndrome.\[1\]

The child had features quite distinctive from DiGeorge syndrome. Chest radiograph showed normal thymic shadow, and echocardiogram and ultrasonogram abdomen were normal. Her severe growth failure and dysmorphic features made us suspect Kenny-Caffey syndrome. Radiograph of long bones were thus taken. These showed cortical thickening with medullary stenosis, and skull X-ray revealed absent diploic space in the skull bones [Figure 2], clinching the diagnosis. Computed tomographic scan of brain was normal.

The child was started on calcium and vitamin D supplements in appropriate doses. She continued to have several admissions with bronchopneumonia, at which time she would develop symptomatic hypocalcemia, suggestive of poor stress response to infection [Table 1]. She required hospitalization and intravenous calcium supplements on each occasion.

In spite of close monitoring and follow-up, the child failed to attain milestones normally and her growth was also slow. At 18 months of age, she developed severe gram-negative sepsis and succumbed to the illness.

**Discussion**

Kenny-Caffey syndrome is a rare hereditary skeletal disorder, first reported by Kenny and Linarelli in 1966.\[2\] Caffey described its radiological features in 1967.\[3\] Lee described the classical facial features in 1983.\[4\]

The children have recognizable identical facies with deep-set eyes, depressed nasal bridge with a beaked nose, long philtrum, thin upper lip, micrognathia, large floppy earlobes, macrocephaly, frontal bossing, delayed closure of anterior fontanel, wide metopic suture and absent diploic space in the skull. The changes in the eyes include microphthalmia, hyperopia, pseudopapilledema, vascular tortuosity, macular clouding, corneal and retinal calcifications and sparse eyebrows and eyelashes. There may be also dental caries, brachymetacarpalia and high hairline. Occasional deafness has been reported. The presenting complaints in all children is hypocalcemic tetany or generalized convulsions detected in the first few days or weeks of life. Occasionally symptoms may be delayed to fourth to seventh month of life.

In addition to the previously described characteristics of the syndrome, hypoplastic nails, persistent neutropenia, abnormal T cell function and neonatal liver disease have been reported by Bergada et al.\[5\] The presence of microorchidism has been reported by Hoffman et al.\[6\]

The radiological evidence of medullary stenosis, osteosclerosis and cortical thickening of long bones was found in most of them. This together with the hypocalcemia, hyperphosphatemia and low concentration of immunoreactive parathyroid hormone in some of them supports the diagnosis. Both autosomal

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**Table 1: Serial calcium, phosphorus and alkaline phosphatase levels**

<table>
<thead>
<tr>
<th>Age in months</th>
<th>Reason for admission</th>
<th>10 m opneumonia</th>
<th>11 m Well</th>
<th>12 m opneumonia</th>
<th>15 m Well</th>
<th>18 m GNB sepsis</th>
</tr>
</thead>
<tbody>
<tr>
<td>Sr. Calcium mg%</td>
<td>Bronch</td>
<td>6.0</td>
<td>9.3</td>
<td>6.1</td>
<td>11.3</td>
<td>7.5</td>
</tr>
<tr>
<td>Sr. Phosphorus (mg %)</td>
<td>Bronch</td>
<td>6.5</td>
<td>5.3</td>
<td>7.1</td>
<td>5.3</td>
<td>6.0</td>
</tr>
<tr>
<td>Alk. Phosphatase (U/L)</td>
<td>Well</td>
<td>266</td>
<td>180</td>
<td>250</td>
<td>169</td>
<td>190</td>
</tr>
</tbody>
</table>
dominant and autosomal recessive inheritance patterns have been described.[7]

The presence of 22q11.2 haploinsufficiency was reported in Kenny-Caffey syndrome, which widens the spectrum of CATCH 22 microdeletion to accommodate Kenny-Caffey syndrome.[8] Karyotyping in this child was normal. The primary outcome of Kenny-Caffey syndrome is short stature. Mental abilities are rarely affected. Mortality of 33% has been reported.[1]

Our child was diagnosed to have Kenny-Caffey syndrome in view of severe growth retardation, typical facial features, hypocalcemia, hypoparathyroidism and characteristic bone changes. Any child with these symptoms, normal cardiovascular examination and intact thymus should raise a suspicion of Kenny-Caffey syndrome. Presence of hypoparathyroidism and typical radiological features are diagnostic. Long-term treatment with calcium and vitamin D supplements is recommended. Outcome may be variable.

References


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