Crossed Polydactyly and Greig Cephalopolysyndactyly Syndrome

R UPPULURI, *K GOWRISHANKAR AND L JANAKIRAMAN

*From Departments of Pediatrics and *Medical Genetics, Kanchi Kamakoti Childs Trust Hospital, 12-A, Nageswara Road, Nungambakkam, Chennai, India.*

Correspondence to: Dr Ramya Uppuluri, House No. 26, 4th Cross Street, Dhandeeswaram Nagar, Velachery, Chennai 600 042, India.
ramya.december@gmail.com
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Greig cephalopolysyndactyly syndrome is a rare genetic disorder, with an autosomal dominant inheritance and consisting of a triad of polysyndactyly, macrocephaly and hypertelorism. Crossed polydactyly is a finding characteristically associated with this syndrome. We report a one and half year old male child who presented with classic clinical features and family history diagnostic of the above syndrome.

Keywords: *Autosomal dominant, Hypertelorism, Macrocephaly, Polysyndactyly.*

Greig cephalopolydactyly syndrome (GCPS) has been described in literature as one of the rare genetic diseases, with an incidence of approximately 1-9/ 1,000,000 [1]. It has not been reported in India till date. GCPS is an autosomal dominant pleiotropic syndrome with multiple congenital anomalies, primarily a triad of polysyndactyly particularly crossed polydactyly, macrocephaly and hypertelorism. We report a case of this extremely rare disorder, wherein a single clinical feature of crossed polydactyly was key to diagnosis.

CASE REPORT

A 1½-year-old boy, born of nonconsanguinous marriage, was admitted with complaints of cough, cold, fever, with no significant past medical or surgical history. On examination, child was stable and had minimal wheeze on respiratory system examination with no organomegaly. On further assessment, child was found to be dysmorphic. He had broad forehead with frontal bossing, broad nasal root, ocular hypertelorism, large head size *i.e.* macrocephaly (head circumference 52cm) which was more than 97th percentile for his age. Crossed polydactyly was noted *i.e.* postaxial polydactyly in his hands and preaxial polysyndactyly of feet. Outward deviation of great toes (delta phalanx) was also noted (***Fig.*1**). His anterior fontanelle was open. He was born at full term, normal vaginal delivery with an uneventful neonatal course. He had attained appropriate motor and mental milestones for his age. Bone age was within normal limits. Echo-cardiography, ultrasound of cranium and abdomen were normal. Family history revealed polydactyly and syndactyly in sister, mother and maternal grandfather, delta phalanx in mother and grandfather, suggestive of an autosomal dominant inheritance. There was no family history of neurological abnormalities.

The above described clinical features with characteristic finding of crossed polydactyly, the positive family history and the autosomal dominant pattern of inheritance are suggestive of Greig cephalopolysyndactyly syndrome. Due to financial constraints, genetic and mutation studies could not be performed.

FIG. 1 *Preaxial polysyndactyly with Delta phalanx .*

DISCUSSION

Crossed polydactyly is defined as presence of preaxial polydactyly of hands and postaxial of feet, or postaxial of hands and preaxial of feet [2]. This finding is found in very few genetic disorders including Ellis van Creveld syndrome. GCPS is characteristically associated with this rare feature [3]. The diagnosis of GCPS requires appropriate clinical features and positive family history. A presumptive diagnosis can be made when a proband has features of preaxial polydactyly, cutaneous syndactyly of toes 1-3 or fingers 3-4, ocular hypertelorism, and macrocephaly [4]. Firm diagnosis requires mutation in *GLI3* gene [5].

The importance of genetic diagnosis is mainly to facilitate early antenatal detection of recurrence in subsequent pregnancies. Management involves a multi-

disciplinary approach including surgical and orthopedic consult for polysyndactyly. Majority of children remain asymptomatic with normal intelligence and activity. Cognitive impairment if present is usually mild. When not associated with neurological abnormalities, children with GCPS can lead a normal life with no decrease in lifespan.

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