Letters to the Editors


To the Editors:

Spondylometaphyseal dysplasia (SMD) is an extremely rare bone dysplasia characterized by vertebral and metaphyseal changes of truncal or juxtatruncal bones of varying severity. Various types of SMD are described with autosomal dominant, autosomal recessive, and X-linked recessive inheritance.1,2 A particular type of SMD termed “cone-rod dystrophy type” has been well characterized by skeletal dysplasia and associated cone-rod dystrophy.3 Ophthalmic abnormalities reported in SMD are retinitis pigmentosa and optic atrophy.4 We report a novel association of SMD with unique form of developmental cataract in two unrelated sibling pairs.

The first sibling pair included 13- and 14-year-old girls who were referred to our institution with a diagnosis of cataract in both eyes for further management. Their physical examination showed short stature with short and stubby fingers and a flat nasal bridge. In the 13 year old, corrected distance visual acuity (CDVA) was hand motions close to face in the right eye and 20/200 in the left eye. Anterior segment examination showed total cataract in the right eye and posterior subcapsular cataract in the left eye. Fundus examination of the left eye was normal. In the 14 year old, CDVA was 20/160 in the right eye and 20/100 in the left eye. Anterior segment examination showed posterior subcapsular cataract in both eyes. Fundus examination was normal. A requested pediatrician consultation showed short-trunk dwarfism with fragmented and flayed metaphyseal ends on x-ray of the knee joint, with no metaphyseal plate thickness in either patient. A diagnosis of SMD with developmental cataract in both eyes was made.

The second sibling pair included 13- and 15-year-old boys who presented to our institution with complaints of diminution of vision in both eyes. Their physical examination showed short stature, genu valgum, short and stubby fingers, and a flat nasal bridge (Figure 1). In the 13 year old, CDVA was 20/200 in the right eye and 20/400 in the left eye. Anterior segment examination showed posterior subcapsular cataract in the right eye and total cataract in the left eye. Fundus examination of the right eye was normal. In the 15 year old, CDVA was 20/100 in both eyes. Anterior segment examination showed posterior subcapsular cataract in both eyes. Fundus examination was normal. Further pediatrician evaluation of the boys revealed SMD on clinical and x-ray examination (Figure 1). A diagnosis of SMD with developmental cataract in both eyes was made.

Electroretinography in all four children was normal, with no evidence of retinal dystrophy. Biochemical tests in both sibling pairs, including serum calcium, phosphorus, total alkaline phosphatase, thyroid profile, and parathyroid hormone E, were normal. The parents were screened and found to have clear lenses in both eyes. A medical examination of all parents revealed no evidence of SMD. Our diagnosis of SMD was made based on radiological and clinical findings. The skeletal phenotype of our patients was identical to those of patients with cone-rod dystrophy type.3 There was no evidence of retinal dystrophy clinically and on electroretinography in any of our cases. There have been reports on association of lens abnormalities

Figure 1. Photographs of the second sibling pair showing (A) short-trunk dwarfism, (B) compressed root of the nose, and (C) genu valgum. Radiographs of (D) hands and (E) knees showing widening of distal metaphyses of the radius and ulna, shortening and widening of short tubular bones of the hand, and metaphyseal flaying with ragged, fragmented metaphyseal ends.
with bone dysplasia such as Stickler and Marshall syndrome, but there are none on developmental cataract associated with SMD in the literature. To the best of our knowledge, this association of developmental cataracts with SMD has never been reported in the literature previously.

REFERENCES

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