

Puberty in the Syndrome of Septo-optic Dysplasia

Hanna et al retrospectively evaluated pubertal development in 13 patients with septo-optic dysplasia. The patients were grouped according to the timing of puberty. Six of the 13 patients comprised group 1; they had clinical signs of puberty beginning earlier than anticipated (bone age <10.5 years in girls, <11.5 years in boys) and experienced rapid progression of puberty associated with bone ages advancing more rapidly than chronologic age. Growth rates were normal-to-increased in this group, but because of the rapid advancement in bone age, these patients lost growth potential. Three of the 13 patients were classified in group 2, with puberty beginning at the expected time and the progression of puberty considered to be normal. The remaining four patients (group 3) were judged to be gonadotropin deficient by low serum levels of follicle-stimulating hormone and luteinizing hormone at bone ages of 12 years in the three girls and 13 years in the boy. Minimal signs of puberty were present at a mean chronologic/bone age of 16.8/13.7 in the girls and 17/13 in the boy, and replacement therapy with sex steroids was instituted.

The authors comment that sexual precocity in girls with septo-optic dysplasia has been described previously. However, they note

that precocity affects boys as well as girls, is most often associated with isolated GH deficiency, and is independent of visual limitation.

Hanna CE, Mandel SH, LaFranchi SH. *Am J Dis Child* 1989;143:186-189.

Editor's comment—*Although this report is a retrospective study, its contributions are important. Twelve of the 13 patients reported in this study received growth hormone therapy. However, only four of these (group 3) had multiple*

hormonal deficiencies and significant pubertal delay or absence. The others either had normal progression of puberty or sexual precocity. Despite the high percentage of patients with abnormal puberty, it is important to note that not all patients with septo-optic dysplasia experience abnormalities of pubertal timing and progression. Thus, it is not possible at this time to make predictions concerning puberty in those children who do not have multiple hormonal deficiencies.

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Birth Prevalence of Skeletal Dysplasias

The prevalence of skeletal dysplasias at birth has received relatively little attention, and the completeness of the available data has been viewed with concern. Two recently reported prospective, population-based studies shed light on this subject.

Stoll and colleagues examined birth records, roentgenographic reports, autopsy reports, follow-up pediatrician notes, and other available data from 11 maternity hospitals where all births were recorded from Strasbourg, France and the surrounding region, from 1979 to 1986. The data from fetuses delivered with a minimum age of 20 weeks and from pregnancies interrupted following prenatal diagnosis of a skeletal dysplasia were included; ascertainment was thought

to be complete. A skeletal dysplasia was diagnosed in 34 cases out of 105,374 births to give a prevalence rate of 32.2 per 100,000. The rates per 100,000 births for several of the more common disorders were: achondroplasia, 6.4; thanatophoric dysplasia, 2.8; achondrogenesis, 2.8; osteogenesis imperfecta, 6.4; osteopetrosis, 1.8; and multiple exostoses, 1.8. Roughly half of the patients had disorders that are usually lethal in the newborn period.

The second study, by Andersen, examined the birth prevalence of lethal bone dysplasias. Clinical and radiographic findings were analyzed from all births, including stillbirths, in the county of Fyn, Denmark, from 1970 to 1983. Twelve