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Picture of the Month

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Denouement and Discussion Acrodysostosis

Manifestations

Acrodysostosis is characterized by peripheral dysostosis, common craniofacial features, skeletal abnormalities, and mental retardation.

The craniofacial abnormalities include brachycephaly, prominent forehead, broad nasal bridge, nasal and maxillary hypoplasia, short upturned nose, apparent ocular hypertelorism, epicanthal folds, delayed eruption of teeth, and hypodontia.

Skeletal abnormalities are the most consistent findings. Peripheral dysostosis, hyperplasia of the big toe, and advanced skeletal maturation occur in most patients. Also present are short limbs, short and broad hands with wrinkling of the dorsal skin, cone-shaped epiphyses that fuse prematurely, and various vertebral abnormalities, the most common one being decreased interpedicular distance. Approximately one third of the patients have urogenital abnormalities, including cryptorchidism, hypoplastic genitalia, and irregular menses.

Approximately 75% of the patients with acrodysostosis are mentally retarded, and 65% have some type of hearing abnormality. Arthritis, scoliosis, and dislocation of the radial heads may develop as the patients become older. Short stature is frequently present.

Genetics

Acrodysostosis usually occurs in a sporadic fashion, although an autosoma-dominant inherited pattern has been reported.

The Editors welcome contributions to Picture of the Month and Radiological Case of the Month. Those who wish to contribute should send their manuscripts to Dr Feingold (Picture of the Month), National Birth Defects Center, Franciscan Children's Hospital, 30 Warren St, Brighton, MA 02135, or Dr Wood (Radiological Case of the Month), Department of Radiology, Childrens Hospital Los Angeles, 1650 Sunset Blvd, Los Angeles, CA 90027. Articles and photographs accepted for publication will bear the contributor's name. There is no charge for reproduction and printing of color illustrations.

Treatment

Treatment consists of management of the skeletal abnormalities, cryptorchidism, hypoplastic genitalia, and mental retardation. Efforts should be directed to help these children achieve their intellectual potential.

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Fig 1 and Fig 2.
Brachycephaly, prominent forehead, broad bridge of an upturned nose, and nasal and maxillary hypoplasia.

Fig 3.
Broad, short hands with excessive skin.

Fig 4.
Roentgenograms of the hands showing dysostosis and cone-shaped epiphyses.