

Arthrogryposis Syndrome (Kuskokwim Disease) in the Eskimo

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A new syndrome of congenital skeletal abnormalities has been found in the Alaskan Eskimo. The condition is characterized by multiple joint contractures predominantly affecting the knees and ankles, with atrophy or compensatory hypertrophy of associated muscle groups. Intellectual functioning is not impaired, and, aside from the skeletal system and possibly the skin, there are no other consistent physical anomalies. General laboratory studies disclosed no abnormality, extensive electromyographic investigation showed no basic defect of the motor unit, and results of muscle biopsies were normal. Treatment to date has been entirely orthopedic, with use of surgery, casting and bracing, and passive manipulation. This condition can be separated from the heterogeneous category of arthrogryposis multiplex congenita by its distinctive clinical features and by the familial pattern of distribution, which suggests an autosomal recessive mode of inheritance.

In the early 1960's, physicians working with the native population of Alaska detected a previously undescribed syndrome of abnormalities of skeletal development. The disorder bears some clinical resemblance to the syndrome of arthrogryposis multiplex congenita (AMC), which is

considered to have multiple causes.¹ Congenital myopathy, anterior horn cell disease, and other conditions which may prevent normal movement either in utero or in the neonatal period are believed to be primarily responsible.²⁻⁴ Most cases occur sporadically, and, although they are frequently classified as either "myopathic" or "neurogenic," findings are often not conclusive. A familial incidence has been reported for the "myopathic" form.⁴ Joint deformity and severe muscle atrophy from disuse have complicated study of this problem, since findings secondary to such changes may be reported as primary to the disease. The disorder found in the Eskimo has unique features which permit its differentiation from the heterogeneous category of AMC. Therefore, we have chosen to call this disorder the "Kuskokwim disease" after the river delta where the original cases were found.

From the beginning, it became apparent that this disorder had a familial distribution and was found only among a group of Southern Eskimos living in a limited geographic area of southwestern Alaska. The syndrome resembles AMC in many respects but differs from cases previously described in that the disorder seems primarily to affect connective tissue.

The total Eskimo population of Alaska numbers about 27,000, of whom some 20,000 are classified by language and cultural characteristics as Southern Eskimos. Of this group, more than half live in the area surrounding the deltas of the Yukon and Kuskokwim rivers. The population of the Kuskokwim delta region is between 10,000 and 12,000, and the 17 verified cases occurred in seven families living in this area.

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