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Metacarpophalangeal Pattern Profile Analysis in Diastrophic Dysplasia

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Abstract

We analyzed the metacarpophalangeal pattern profile (MCP) on 16 individuals with diastrophic dysplasia and calculated a mean syndrome profile. Correlation studies confirm clinical homogeneity of the hand profile in diastrophic dysplasia. Discriminant analysis of individuals with diastrophic dysplasia compared with a sample of normal individuals produced a function of 3 MCP variables plus age that appears to be a useful diagnostic tool.

Keywords

MCP; discriminant analysis; correlation studies; autosomal recessive inheritance

INTRODUCTION

Diastrophic dysplasia (DD) was characterized by Lamy and Maroteaux in 1960. The patients are short of stature and have short tubular bones, limitation of joint motion with talipes, scoliosis, hypertrophied auricular cartilage, and normal intelligence [Smith, 1982]. DD is an autosomal recessive trait. Due to phenotypic variability, early diagnosis may be difficult. Therefore, quantitative methods based on radiographic measurements may be helpful [Poznanski, 1984].

Metacarpophalangeal pattern profile (MCP) analysis is an evaluation of the hand skeleton based on a comparison of the 19 tubular bone lengths to normal bone-length standards, as described by Poznanski et al [1972] and Garn et al [1972]. This method provides a quantitative assessment of the amount and direction of abnormality of the hand skeleton. MCP analysis has been used to evaluate numerous syndromes [Poznanski, 1984; Butler et al, 1986].

Recently we derived a method of MCP analysis for 16 individuals with DD to evaluate its potential as a diagnostic technique.

MATERIALS AND METHODS

MCPP Data

Postero-anterior hand radiographs were obtained on 16 individuals diagnosed with DD. The diagnosis was made by at least 2 physicians on more than one occasion. The patient group included 9 males and 7 females ranging in age from $1\frac{1}{12}$ to 38 years, with a mean age of $12\frac{11}{12}$ years.

The length of each metacarpal and phalangeal bone of each patient was measured in millimeters with a vernier caliper and compared to bone-length standards (appropriate for age and sex) published by Garn et al [1972] (white Americans, age 2 years to adulthood) and Poznanski [1974] (Gefferth Hungarian sample, birth to 15 months). Through these comparisons, Z score values for the 19 bones of each patient were obtained ($Z \text{ score} = \text{observed bone length} - \text{mean bone length} \div \text{SD}$). Therefore, MCPP on a given patient is a set of 19 Z scores, which may be plotted on a graph or subjected to various statistical procedures for study and comparison with the MCPP of other patients or groups of patients [Poznanski et al, 1972].

Correlation Studies

We derived a mean pattern profile, based on the average Z score for each bone, from the 16 patients [Poznanski et al, 1972; Garn et al, 1972]. The pattern for each patient was compared to this group mean pattern and to each other using Pearsonian correlation coefficients.

Discriminant Analysis

A forward stepwise method of discriminant analysis [Enslein et al, 1977] was performed on the 19 Z score variables and age of individuals from 2 groups: the 16 patients with DD and a control group of 41 normal individuals whose hand radiographs were randomly obtained from the records of Indiana University School of Dentistry. The 41 normal individuals included 17 males and 24 females, with an age range of $9\frac{6}{12}$ to 18 years and a mean age equal to $13\frac{1}{12}$ years.

RESULTS

The mean Z scores fall between -2.2 and -5.4 . Therefore, each measured hand bone is significantly shorter than the mean of normal individuals with no apparent overlap between DD and normal. The mean pattern profile based on the 16 patients with DD contains 2 prominent peaks (first proximal and second distal phalanges) (Fig. 1).

Next, the correlation program was used to assess similarity between the mean pattern and each of the 16 individual patterns. Twelve of 16 individuals have significant positive correlations (Table I).

Discriminant analysis of the normal and DD cases resulted in a discriminant function based on 3 of the 19 MCPP variables and age. In the discriminant analysis, patients with DD were distinguished from normal individuals at an overall correct classification rate of 100% for this sample (Fig. 2). The 3 MCPP variables in the discriminant function were the Z scores

representing the third metacarpal (X3), the fifth metacarpal (X5), and the second middle phalanx (X11).

DISCUSSION

Small hand size is a characteristic of DD individuals. The mean pattern profile based on our 16 DD patients confirms this in quantitative terms. All the digits are short with marked shortness of the metacarpals, particularly on the radial side. The shortest bone relative to normal is the first metacarpal, while the proximal phalanx of the thumb is relatively long. The correlations with the DD individuals suggest a homogeneous pattern, with 75% of the individuals possessing a significant correlation with the Z score group mean. Therefore, a unique hand profile exists in DD based on these measurements.

The results from the discriminant analysis suggest that effective diagnosis of DD is possible on the basis of MCP data. We are encouraged by these results, especially since the hand films of 5 individuals were studied at or below age one year. Additional testing with more individuals is needed to test the power of the discriminant method to distinguish DD patients not only from normal individuals but from patients with other conditions with small hands and/or a generally similar phenotype including the DD variant. The observations presented in this report suggest the potential of MCP analysis as a diagnostic tool in the evaluation of patients in whom DD is considered.

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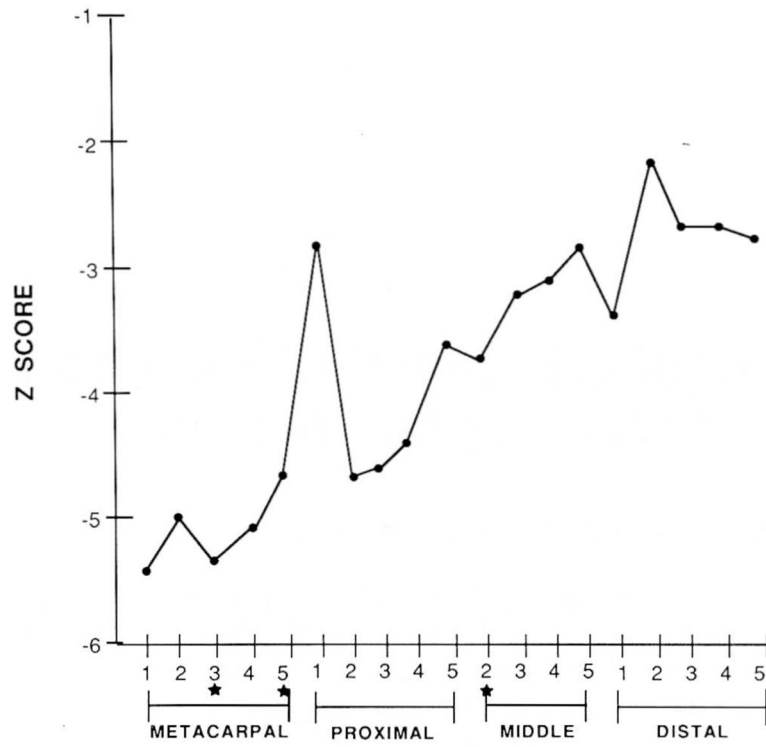


Fig. 1. Mean MCPP of 16 individuals with diastrophic dysplasia. ★, Bones that were selected in the discriminant analysis.

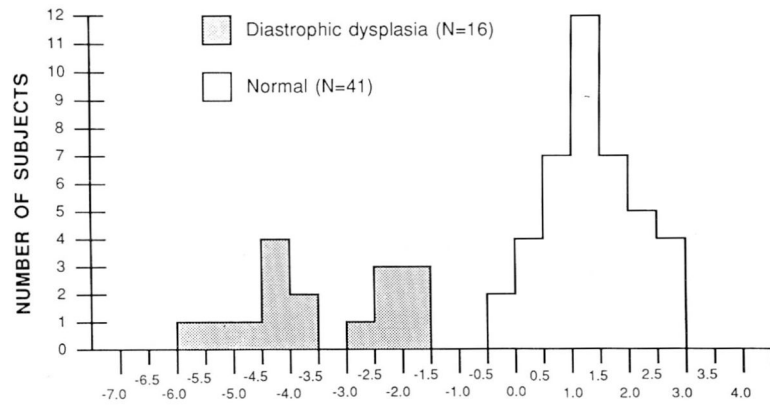


Fig. 2. Histogram depicting normal and diastrophic dysplasia classification by discriminant analysis. $D = -0.50 + 1.26 (X3) - 0.73 (X5) + 0.37 (X11) + 0.14$ (age in years, 18 if an adult).

TABLE I

Correlation Between Individual and Group Mean MCPP in Diastrophic Dysplasia

Age (yr)	Sex	Correlation
1.1	M	0.75 ^a
4.0	M	0.53 ^a
7.0	M	0.42 ^b
15.0	M	0.84 ^a
16.0	M	0.76 ^a
16.0	M	0.65 ^a
18.0	M	0.14
25.0	M	0.35
36.0	M	0.76 ^a
0.1	F	0.52 ^b
0.2	F	0.24
0.2	F	0.88 ^a
0.3	F	0.75 ^a
2.1	F	0.71 ^a
27.0	F	0.61 ^a
38.0	F	0.08

^aP < .05 for 1-tailed test.^bP < .005 for 1-tailed test.