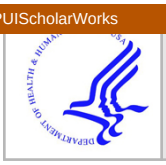

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Metacarpophalangeal Pattern Profile Analysis in Robinow Syndrome

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Abstract

We analyzed the metacarpophalangeal pattern profile (MCPP) on 15 individuals with Robinow syndrome and calculated a mean Robinow syndrome profile. Correlation studies confirm clinical homogeneity of the hand profile in the Robinow syndrome. Discriminant analysis of individuals with Robinow syndrome compared with a sample of normal individuals produces a function of 6 MCPP variables that may provide a useful tool for diagnosis.

Keywords

Robinow syndrome; fetal face syndrome; metacarpophalangeal pattern profile (MCPP); discriminant analysis; correlation studies

INTRODUCTION

The Robinow syndrome or “fetal face” syndrome was first described by Robinow et al [1969]. More than 30 cases were reported subsequently [Butler and Wadlington, 1987]. Affected patients have short forearms and hands, short stature, flat facial profile with hypertelorism, a short upturned nose, vertebral abnormalities and hypoplastic genitalia. Most cases are sporadic, although autosomal recessive [Wadlington et al, 1973; Seemanová et al, 1974; Wadia et al, 1978; Saal et al, 1985] and autosomal dominant [Robinow et al, 1969; Shprintzen et al, 1982; Vallee et al, 1982] inheritance patterns have been reported. Due to considerable phenotypic variability, early diagnosis may be difficult, especially in females and without other affected relatives. Therefore, quantitative methods based on radiographic measurements may be helpful as suggested by others [Giedion et al, 1975; Robinow and Chumlea, 1982].

Metacarpophalangeal pattern profile (MCPP) analysis is an evaluation of the hand skeleton based on a comparison of the 19 tubular bone lengths to normal bone-length standards, as

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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. MCPP analysis has been used to evaluate numerous syndromes [Poznanski, 1984; Butler et al, 1986].

Recently we derived a method of MCPP analysis for 15 individuals diagnosed with Robinow syndrome to evaluate its potential as an additional diagnostic technique.

MATERIALS AND METHODS

MCPP Data

Postero-anterior hand radiographs were obtained on 15 individuals diagnosed with Robinow syndrome. The diagnosis was made by at least 2 physicians on more than one occasion. The patient group included 11 males and 4 females ranging in age from 8/12 to 13 8/12 years, with a mean age of 11 7/12 years.

The metacarpophalangeal bone lengths of each patient were 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 score = observed bone length − mean bone length ÷ 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 15 patients [Poznanski et al, 1972; Garn et al, 1972]. The pattern from 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 15 patients with Robinow syndrome 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 6/12 to 18 years and a mean age equal to 13 1/12 years.

RESULTS

The mean Z scores fall between −2.0 and −4.1. Therefore, each measured hand bone is significantly shorter than the mean for normal individuals with no apparent overlap between Robinow syndrome and normal. The mean pattern profile based on the 15 patients with Robinow syndrome contains 3 peaks (proximal, middle and distal phalanges, Fig. 1). The

longest bone is the first proximal phalanx while the shortest bone is the fourth middle phalanx.

Next, the correlation program was used to assess similarity between the mean pattern and each of the 15 individual patterns. Thirteen of 15 individuals have significant positive correlations (Table I).

Discriminant analysis of the normal and Robinow syndrome cases resulted in a discriminant function based on 6 of the 19 MCPP variables. In the discriminant analysis, patients with Robinow syndrome were distinguished from the normal individuals at an overall correct classification rate of 98% for our sample (Fig. 2). The 6 MCPP variables in the discriminant function were the Z scores representing the third metacarpal (X3); the second (X11), fourth (X13) and fifth (X14) mid phalanges; the second (X16) and fourth (X18) distal phalanges.

DISCUSSION

Small hand size is a recognized characteristic of individuals with the Robinow syndrome. The mean pattern profile based on our 15 patients confirms this characteristic in selected quantitative terms. The correlations with the Robinow syndrome individuals suggest a homogeneous pattern with 87% of the individuals possessing a significant correlation with the mean group. There appeared to be no hand profile differences between sporadic or familial cases. Therefore, a unique hand profile exists for Robinow syndrome based on these measurements.

The results from the discriminant analysis suggest that effective delineation of Robinow syndrome patients from normal individuals is possible on the basis of MCPP data. We are encouraged by these results, especially since the hand films of several individuals were studied at a young age. Additional testing with a larger sample size is needed to test the power of the discriminant method to distinguish patients with Robinow syndrome not only from a normal sample but from patients with other conditions with small hands and/or a generally similar phenotype. The observations presented in this report suggest the potential of MCPP analysis as a diagnostic tool in the evaluation of patients in whom Robinow syndrome is considered.

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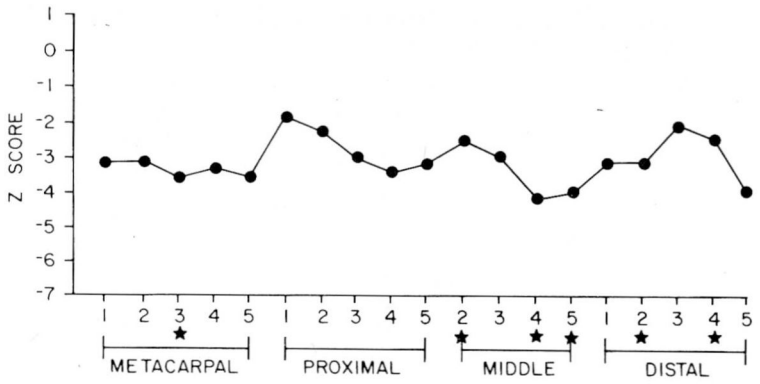

Fig. 1.

Mean MCPP for 15 individuals with Robinow syndrome. ★, Bones that were selected in the discriminant analysis.

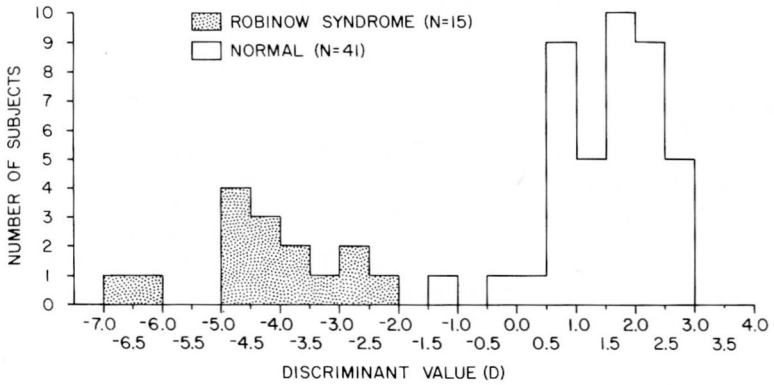

Fig. 2.

Histogram depicting normal and Robinow syndrome classification by discriminant analysis. D = 1.13 + 0.56 (X3) − 0.94 (X11) + 0.76 (X13) + 0.60 (X14) + 0.68 (X16) − 0.75 (X18).

TABLE I

Correlations Between Robinow Syndrome Individual's MCPP and Group Mean MCPP

Age (years)	Sex	Correlation
0.8	M	0.19
2.1	M	0.47[a]
2.5	M	0.42[a]
3.0	M	0.60[a]
7.8	M	0.78[a]
9.3	M	0.80[b]
12.7	M	0.55[a]
16.0	M	0.45[a]
18.3	M	0.64[b]
21.8	M	0.65[b]
23.0	M	0.66[b]
6.0	F	0.47[a]
15.2	F	0.51[a]
16.1	F	0.36
20.3	F	0.66[b]

[a] $P < .05$ for one-tailed test.

[b] $P < .005$ for one-tailed test.