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PRELIMINARY COMMUNICATION: PHOTOANTHROPOMETRIC ANALYSIS OF INDIVIDUALS WITH THE FRAGILE X SYNDROME

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Abstract

A photoanthropometric method, which enables an objective description of facial structures, was used to examine 31 boys with the fragile X or Martin-Bell syndrome below the age of 12 years. The age range was 1.5 to 12 years with an average age of 6.5 years. Facial parameters were measured from strict frontal and profile photographs of fra(X) syndrome boys and compared with other facial measurements from the same face (e.g. mouth width vs. bizygomatic diameter). We studied 18 photoanthropometric facial parameters following the protocol established by Stengel-Rutkowski et al [1984]. Fourteen indices were calculated and compared with photoanthropometric index standards for age, established from normal children between 0 and 12 years [Stengel-Rutkowski et al, 1984]. Two of the fourteen craniofacial indices, broad palpebral fissures and decreased inner canthal distance, were significantly abnormal.

Keywords

Photoanthropometric parameters; fragile X syndrome; broad palpebral fissures

INTRODUCTION AND METHODS

Although the fragile X (or Martin-Bell) syndrome is a well - recognized condition and the most common cause of familial mental retardation, few anthropometric studies are available [Brown et al, 1981; Meryash et al, 1984; Partington, 1984]. Therefore, we used a photoanthropometric method, which enables an objective description of facial structures, to examine 31 boys with the fra(X) syndrome below the age of 12 years. The age range was 1.5 to 12 years with an average age of 6.5 years. The average fra(X) chromosome expression was 23% with a range of 3 to 60%. We studied 18 facial parameters (inner canthal distance; palpebral fissure width; bizygomatic diameter; midface height; mouth width; nose length; interalar distance; trignon-subnasale, trignon-ophryon, and trignon-chin distances; ear length and width; conchae length and width; nasolabial distance; chin and total face heights; and

ear position) following the protocol established by Stengel-Rutkowski et al [1984]. Facial parameters were measured to the nearest 0.1 mm with a vernier caliper from strict frontal and profile photographs obtained from a distance of over 1.5 m from fra(X) syndrome boys and compared with other facial measurements from the same face (e.g. mouth width vs. bizygomatic diameter). Fourteen indices were calculated and compared with photoanthropometric index standards for age, established from normal children between 0 and 12 years [Stengel-Rutkowski et al, 1984]. Herein, we report data from several facial photoanthropometric parameters of individuals with the fra(X) syndrome. Additional parameters will be reported at a later date.

RESULTS

Our data on 14 craniofacial photoanthropometric indices indicated that 2 of 4 facial indices were significantly abnormal. These included: palpebral fissure width to bizygomatic diameter [23 of 28 individuals or 82% were 97th centile; correlation coefficient (r) with age = -0.24 ($p > 0.05$)] with an average Z score of 3.3; and, interalar (nose width) distance to bizygomatic diameter [19 of 29 individuals or 66% were 97th centile; correlation coefficient (r) with age = -0.03 ($p > 0.05$)] with an average Z score of 2.2. Mouth width to bizygomatic diameter [average Z score of 1.6; correlation coefficient (r) with age = -0.03 ($p > 0.05$)] and inner canthal distance to bizygomatic diameter [average Z score of 0.4; correlation coefficient (r) with age = -0.20 ($p > 0.05$)] were not significantly abnormal. Therefore, the face was either narrow with a decreased bizygomatic diameter and/or there was increased palpebral fissure width and interalar distance. The impressions of a narrow face, broad nose and broad palpebral fissures are supported by our data as shown in Fig. 1. The inner canthal distance to bizygomatic diameter was normal (average Z score of 0.4) which would also suggest that the inner canthal distance may be decreased although the apparent narrow inner canthal distance may be due to broad palpebral fissures.

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SUMMARY

Thus, our photoanthropometric analysis supports the clinical impression of a narrow face with a broad nose and broad (greatest distance of horizontal opening of the eye) palpebral fissures. There were no significant correlations between age and the 4 facial photoanthropometric indices. Although a narrow face and a broad nose are not new manifestations, broad palpebral fissures were previously unrecognized and undocumented in individuals with the fra(X) syndrome. These photoanthropometric parameters may be useful in identifying individuals for fra(X) chromosome analysis, although more research is needed to better define the craniofacial structures and changes in individuals with the fra(X) syndrome.

Figure 1.
Four facial indices (modified from Stengel-Rutkowski et al, 1984) showing photoanthropometric data collected from fragile X syndrome individuals.