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Trisomy 5p syndrome

Sir,

DiLiberti et al. (1977) suggested that trisomy 5p has a clinically recognisable and consistent phenotype. In their report on 'Complete trisomy 5p', Brimblecombe et al. (1977) suggest that the phenotype in trisomy 5p is non-specific. I have also found three cases (males aged 22, 23, and 7 years) in a family with a partial trisomy 5p13-->pter, and each of these patients had the following features: a full face with the appearance of jowls, a long philtrum, convergent strabismus, a bulbous nose, mild malar hypoplasia, prognathism, obesity, and mental retardation. The dermatoglyphs showed a characteristic combination of patterns: an excess of ulnar loops on the fingertips, transversal direction of the dermal ridges on the palms, t' triradii, a unilateral, atypical simian crease, and tibial arches on the halluca areas. A high total finger ridge count was found in only one case (case 3, TFRC = 167; mean value in the normal male population, X = 145, Rodewald et al., 1977; case 1, TFRC = 102; case 2, TFRC = 100).

Several of the dermatoglyphic manifestations in the 5p+ syndrome are strikingly different from features described in cases with partial monosomy 5p-; a high frequency of whorls on the fingertips, longitudinal direction of the dermal ridges on the palms, t' or t' triradii or both, a typical simian crease, and distal loops on the hallucal areas. This contrast confirms, in the context of chromosome 5, the hypothesis of a 'contre type' that Lejeune et al. (1964) proposed for the clinical picture of trisomy 21 (Down's syndrome) and its counter-type, monosomy 21.

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References

Single crease on the 5th finger in medical disorders and in normal population

Sir,

In their recent report on 'Clinical experience with trisomies 18 and 13', Hodes et al. (1978) found a single flexion crease on digit 5 in 63% (20 palms in 32 patients) of trisomy 18 patients and in only 5% (1 palm in 19 patients) in trisomy 13 patients. Penrose (1931) first described the presence of a single crease on digit 5 in 16 of 60 cases of Down's syndrome (26.7%). In 8 of these, the single crease was present on one hand only. We found this peculiarity in a group of 200 patients with Down's syndrome (trisomy 21), in 21.5% symmetrically and in 4.5% asymmetrically. The presence of the single crease on both hands (symmetrical) was found in 26.4% of the male patients and only in 13.4% of the female patients. This pattern has also been described in about 19% of patients with trisomy 18 by Schaumann and Alter (1976), in 43.2% (44 palms) of patients with partial trisomy 9p by Rodewald (1979), and occasionally in partial monosomy 18q (Lejeune et al., 1966), in de Lange's syndrome (Pfeiffer and Kumbhani, 1966; Smith, 1966), and in other medical disorders (Schaumann and Alter, 1976). Cumming and Midlo (1961) noted the absence of the distal interphalangeal flexion crease on an immovable distal joint on the left ring finger of a Negro boy. We found a single crease on the 5th finger in two patients with partial trisomy 9p but not in patients with the trisomy 8 syndrome (Rodewald et al., 1977) or patients with partial trisomy 10p (Rodewald and Stengel-Rutkowski, 1978).

So far, a single crease on the 5th finger in a phenotype typically and chromosomally normal subject has not been reported. However, we found a single crease on the 5th right finger, without aplasia of the middle phalanx and with a movable interphalangeal joint, in a phenotypically and chromosomally normal man (Fig).

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