Correspondence

Received: March 15, 1983

Accepted: May 25, 1983

Acta haemat. 1983;70:275-276

The Pelger-Huet Anomaly: A New Familial Association with Polydactyly and Trisomy 13 Syndrome

M.	Mikhail	Berman
W.	Walter	Fried
W.	William	Knospe

Chicago

Mikhail Berman, Walter Fried, William Knospe, Section of Haematology, Department of Internal Medicine, Rush Presbyterian St. Luke's Medical Center, Chicago, IL 60612 (USA)

The Pelger-Huet (PH) anomaly is an autosomal dominant hereditary trait characterized in the heterozygous state by increased condensation of nuclear chromatin, poor segmentation of granulocytes, and a benign course. Its unique nuclear phenotype suggests an inherited defect in the genetic control of the last postmitotic stage of granulocytic maturation which begins with elongation of nucleus of metamyelocyte. So far no studies have shown any correlation between PH and the specific autosome that carries mutant gene or genes. In one study, linkage was established between PH and an unusual autosomal form of muscular dystrophy [1]. Unfortunately, the latter trait has no chromosomal assignment.

In 1981, Aznar and Vaya [2] described an 18-month-old girl with the homozygous form of PH and 6 fingers on one hand and 6 toes on both feet. Poly-

dactyly also was observed in the propositus' sister and in a first cousin of the maternal grandmother.

We have observed a black family in which the father and 2 sons had the heterozygous form of PH. The affected 3 ½-year-old boy had an additional digit without bone structure attached to his right thumb. 1 of 2 daughters in this family died in 1972 of multiple

congenital malformations at age 3 months. She had microcephaly, a defect over the posterior region of the head through which meninges were visible, hy-pertelorism, microphthalmia, low-set ears, a small posterior cleft in the palate, and a simian crease. An extra digit was attached to the fifth finger and fifth toe on both hands and feet. Her hemoglobin was 13.5, hematocrit 40.9, white blood cell count 9,000, with 40% segments, 1 % bands, 45% lymphocytes, 1 % eosinophils and 13% monocytes. The skull X-ray

276

Correspondence

showed lack of ossification of the parietal bone posteriorly in the midline consistent with cranium bifi-dum.

The infant was discharged at about 2 months of age in fair condition. She died at home 1 month later of complications of her severe multiple malformations.

A chromosomal study confirmed the clinical impression of trisomy 13 syndrome with unbanded kar-yotype 46, XX,-D,+t(Dq;Dq). The other family members had normal karyotypes.

Several skeletal malformations have been described in rabbits with the homozygous form of PH [3]. Based on this model, Nachtsheim [3] predicted similar bone abnormalities in homozygous humans and his predictions have now turned out to be at least partially correct.

Peculiar nuclear projections have been observed in the granulocytes in more than 50% of patients with trisomy 13 syndrome. The locus responsible for this anomaly was tentatively assigned to region located close the centromere on the proximal portion of the long arm of chromosome 13 on bands 13q12 and the uppermost part of 13q13 [4]. Polydactyly occurs in the trisomy 13 syndrome more often than in any other congenital anomaly. It is linked to the distal portion of the long arm of chromosome 13 on bands 13q31-13q34[5].

It is premature to draw conclusions about the chromosomal assignment of the PH anomaly. Appropriate linkage studies between a given anomaly and traits or markers positively assigned to chromosome 13 could be done to evaluate the significance of this new familial association of the PH anomaly and polydactyly.

Acknowledgements

The authors appreciate discussions with Daniel L. Van Dyke, PhD, Director, Cytogenetics Laboratory, Henry Ford Hospital, Detroit, Mich.

References

Schneiderman, L.J.; Sampson, W.I.; Schoene, W.C.; Haydon, G.B.: Genetic studies of a family with two unusual autosomal dominant conditions: muscular dystrophy and Pelger-Huet anomaly. *Am. J. Med.* 46: 380-393 (1969).

Aznar, J.; Vaya, A.: Homozygous form of Pelger-Huet anomaly in man. *Acta haemat.* 66: 59-62 (1981).

Nachtsheim, H.: The Pelger-anomaly in man and rabbit. *J.Hered.* 41: 131-138(1950).

Huehns, E.R.; Lutzer, M.; Hecht, F.: Nuclear abnormalities of the neutrophils in a D₁₃ (13-15) trisomy syndrome. *Lancet* i: 569 (1964).

Escobar, J.I.; Yunis, J.J.: Trisomy for the proximal segment of the long arm chromosome 13. *Am. J. Dis. Child.* 128: 221-222(1974).

)

Acta haemat. 70: 276-277(1983)

Platelet Function in Dengue Hemorrhagic Fever

To the Editors,

In dengue hemorrhagic fever (DHF), the bleeding diathesis is an important part of the disease. The

nature of the hemostatic defect has not been completely defined, however. It was suggested that three mechanisms are involved; vascular injury, coagulo-pathy and thrombocytopenia [1]. On the other hand,