



## Homozygous form of the Pelger-Huët anomaly

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**A** case of homozygous Pelger-Huët anomaly is reported, showing extreme hypolobulation of peripheral blood cells and giant platelets. After fifteen years of follow-up, only slight neutropenia and thrombocytopenia have appeared in this otherwise healthy patient.

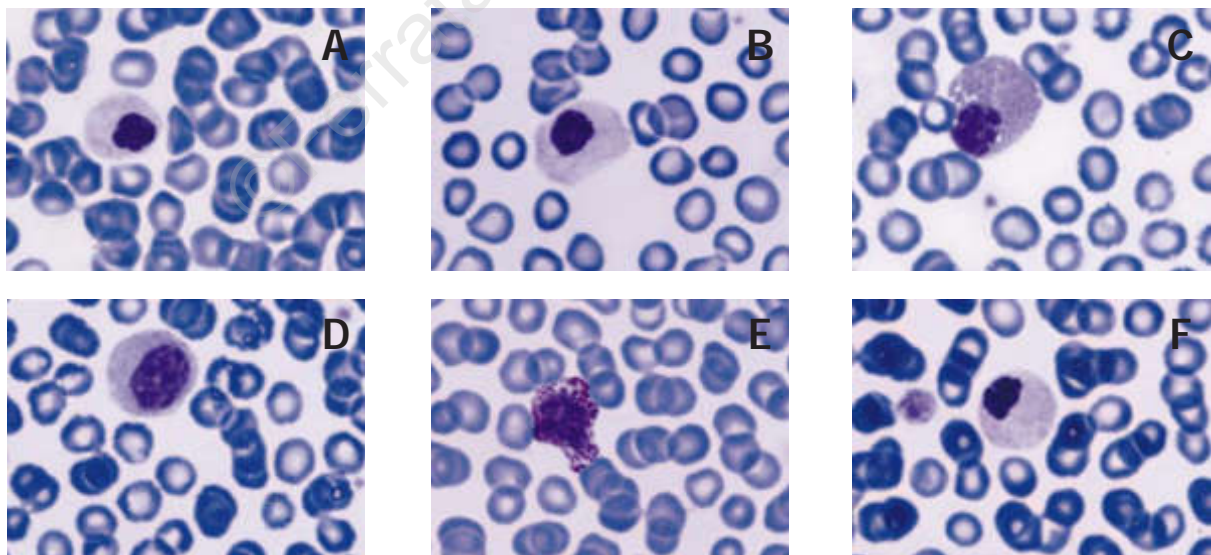
The Pelger-Huët anomaly, first described in 1928, is a well-known morphologic leukocyte anomaly in which the nuclear lobes fail to develop completely. This must be borne in mind to avoid a false diagnosis of *left shift*. The inheritance is autosomal dominant, with an incidence about 1/5,000.<sup>1,2</sup> The anomaly has occasionally been described in association with other syndromes.<sup>3</sup> Acquired forms can be seen associated with myelodysplastic and leukemic conditions. Homozygous forms are very rare, and fewer than ten cases have been reported to date.<sup>1</sup> The paucity of reports in spite of the high incidence of the heterozygous form, as well as results of studies carried out in rabbits, suggest that the homozygous form might be lethal.<sup>4</sup>

The patient reported here is a 30-year old male first diagnosed in 1982, when both parents were found to carry the anomaly.<sup>5</sup> To date, only a dorsal hyperkyphosis, short stature, slightly short upper limbs, and dyshidrosis, are of note among clinical findings. Complete blood count shows a slight neu-

tropenia (1,400 granulocytes/ $\mu$ L) and thrombocytopenia (130,000 platelets/ $\mu$ L), the other parameters being within normal limits. In the blood smear all granulocytes have a round nucleus with coarsely clumped chromatin, while cytoplasm appears mature (Figure 1, A, B, F). This failure to develop lobulation is found to affect eosinophil, basophil and monocytic lineages as well (Figure 1, C, D, E). Some platelets are giant, sometimes larger than an erythrocyte (Figure 1, F).

### References

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**Figure 1.** A and B: Mature neutrophils showing extreme hypolobulation and coarsely clumped chromatin. C: Non-lobed eosinophil. D: Monocyte with oval nucleus. E: Non-lobed basophil. F: Giant platelet next to a mature neutrophil. (May-Grünwald-Giemsa, x 1,000).

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