Familial Thrombocythemia and/or Thrombocytosis – Apparently a Rare Disorder

M.L.		Randi
F.		Fabris
C.		Vio
A.		Girolami

Padua

M.L. Randi, F. Fabris, C. Vio, A. Girolami, University of Padua, Institute of Medical Semeiotics, Via Ospedale, 105, I-35100 Padua (Italy)

Several instances of familial polycythemia vera (PV) have been described [1,6, 9]; however, no data are available on the incidence of primary or secondary thrombocytosis in the same family. Over the last 10 years we have studied 170 subjects with thrombocytosis. 133 of them (65 males and 68 females) presented a primary form: 63 essential thrombocythemia (ET), 52 PV, 5 myelofibrosis with myeloid metaplasia (MMM) and 13 chronic myeloid leukemia (CML). The other patients (37 cases, 16 males and 21 females) showed a secondary thrombocytosis due to neoplasms, miscellaneous chronic conditions or splenectomy. The platelet count was performed in EDTA blood by phase microscope using the Un-opette diluting system (Beckton-Dickinson, USA). Platelet aggregation was performed as previously described [2] under stimuli with ADP 2 μ M, adrenalin 10 μ M and collagen 2 μ g. The platelet aggregation was expressed as normal, increased or decreased in comparison to 100 controls comparable for sex and age. Platelet serotonin content (5-HT) was evaluated by a modification of the method proposed by Udenfriend as previously described [3] (normal value: 1.5–3 nM/109 platelets).

In the primary thrombocytosis group we have found only 2 related subjects (1.5%) out of 133 patients. They were 2 sisters, one with PV (patient 1) and the other with an ET (patient 2). For the diagnosis we accepted the criteria proposed by Polycythemia Vera Study Group [1]. The main features of the 2 selected patients are summarized in the following table:

Patient 1
Patient 2
Age at diagnosis
56
53
Duration of disease, years
5
1
Thromb/hemorrh.
no
no

Platelets, $\times 10^9$

540

859

Aggregation

ADP

decreased

increased

Adrenalin

decreased

normal

Collagen

decreased

normal

5-HT, ng/ 10^9 platelets

1.33

1.5

In the cases of secondary thrombocytosis no familial forms were found. The fact that a sister presented a typical PV and the other an ET is of no surprise. It is well-known that the different myeloproliferative disorders are maintained to be due to an aberration of a bone marrow stem cell. The mechanism of thrombocytosis in response to different stimuli is probably individually motivated as indicated by the negative results observed in the secondary forms. Primary forms of familial thrombocytosis also seem to be a rare disorder. On the contrary, familial thrombocytopenia is much more frequent [7, 8], both without or with morphologic changes as in May-Hegglin anomaly [4, 5]. This discrepancy remains unexplained.

References

- Brubaker, L.H.; Wasserman, L.R.; Goldberg, J.D.; Pisciotta, A.V.; McIntyre, L.; Kaplan, M.E.; Modan, B.; Flannery, J.; Harp, H.: Increased prevalence of polycythemia vera in parents of patients on polycythemia vera study group protocols. *Am. J. Hematol.* 16: 367–373 (1984).
- Fabris, F.; Randi, M.L.; Sbrojavacca, R.; Casonato, A.; Girolami, A.: The possible value of platelet aggregation studies in patients with increased platelet number. *Blut* 43: 279–285 (1981).
- Fabris, F.; Randi, M.L.; Casonato, A.; Dal Bo Zanon, R.; Bonvicini, P.; Girolami, A.: Clinical significance of beta-thrombo-globulin patients with high platelet count. *Acta haemat.* 7: 343–346 (1984).
- Girolami, A.; Randi, M.L.; Casonato, A.; Pasini, L.; Boccato, C.; Fabris, F.: A study of platelet function and morphology in a new family with May-Hegglin anomaly. *Folia haemat.* 707: 256–268 (1980).
- May, R.: Leukocyteninschlüsse. *Arch. klin. Med.* 96: 1–12 (1909).
- Modan, B.: Polycythemia. A review of epidemiological and clinical aspects. *J. chron. Dis.* 18: 605–645 (1965).
- Schaar, F.E.: Familial idiopathic thrombocytopenic purpura. *J. Pediat.* 8: 397–399 (1963).
- Schainson, G.: Thrombopénie familiale chronique. *Nouv. Revue fr. hémat.* 62: 546–551 (1968).
- Weber F.P.: The constitutional and familial factor in primary and secondary polycythemia rubra. *Med. Press* 724: 128–133 (1927).