Clinical review

Recent advances in haematology

Drew Provan, Denise F O'Shaughnessy

Haematology is a diverse specialty embracing clinical and laboratory aspects of adult and paediatric disease, both malignant and non-malignant. We describe here some of the progress that has been made in diagnostic and therapeutic strategies. Molecular advances are continuing at a phenomenal rate (exceeding the rate of progress in therapeutics), offering highly sensitive methods for disease detection and, in some disorders, prospects of our ethnough gene theraps.

Methods

We used information from recent key meetings, including those of the British Society for Elaematology and the American Society of Hermatology; leading articles in major harmatology journals; and discussion with colleagues. The choice of topics covered is largely personal, and osting to space restrictions we have not included every "advance" within the specialty.

Amaginalias

Distinguishing between anaemia due to iron deficiency and anaemia of chronic disease is a difficult but common problem in medical practice. Anaemia of chronic disease is complex and involves inflammatory extokines," reduced marrow response to erythropoietin, reduced red cell life span, and impaired reuse of iron.1 In typical, uncomplicated iron deficiency anaemia the haemoglobin concentration, mean cell volume, and serum ferritin and iron concentrations are reduced with raised total iron binding capacity; unforturnately cases are often not typical, and the results of these tests may seem conflicting. Furthermore, if a patient with amarmia of chronic disease is also iron deficient, the parameters for diagnosing iron deficiency are altered, making the diagnosis difficult; often a borse marrow aspirate, stained for iron, is the only method for accurately assessing from status. This is expensive, time consuming, and unpleasant for the patient. Recently the serum transferrin receptor assay has been developed, enabling more accurate assessment of iron status in this group of patients,

Serum ferritin, a 480 kDa multisubunit protein, represents the body's iron storage pool. A reduced serum ferritin concentration generally indicates depletion of the iron stores Ferritin, however, is an "acute phase protein," whose concentration is raised in inflammatory disorders, in a patient with, for example, active rheumatoid disease the ferritin concentration may be normal (or even raised) even if the patient is

Recent advances

from deficiency arusemia can more be distinguished from arusemia of chronic disease by using serum transferrin receptor assays

A recently discovered human herpesvirus may have a role in development of raveloma

Additional thrombotic risk factors—for example, factor V and prothrombin gene mutations, hyperhomocysteinaemia—have been identified

Newly designed DNA probe arrays have the potential to allow rapid automated detection of mutations

Antisense therapy, ribosyme technology, and other novel treatment methods are being developed

truly iron deficient. The transferrin receptor assay aims to distinguish clearly between simple iron deficiency and anaemia of chronic disease in most cases.

How does the transferrin receptor away work? Transferrin is a protein that carries plasma iron. Cellsthat require iron express the transferrin receptor on the cell surface." The receptor binds plasma transferrin with bound iron atoms, after which the transferrin receptortransferrin complex is internalised (fig 1). After unloading the iron atoms, the transferrin receptor-transferrin complex moves back to the cell surface and dissociates, leaving transferrin free to pick up further iron atoms, In iron deficiency anaemia the number of transferrin receptors increases (is upregulated)," whereas in anacmia of chronic disease the number of transferring receptors remains normal. Assays for transferrin receptors may be performed on serum with an enzyme linked irruntinosorficut assoc (ELISA), allowing large numbers of samples to be batch tested. According to available data, the test seems as reliable as testing bone marrow assistate for diagnosing from deficiency and should be valuable in potients with riscurratology disorders and others with inflammatory disorders.

Haemophilia

Haemophilias A (factor VIII deficiency) and B (factor IX deficiency, Christmas disease) are X linked blooding

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de Loo, J. Ritter, J. Kienast, J. Vormoor, 2012-12-06 Some three decades after bone marrow transplantation was introduced in the field of hematology and oncology transplantation today continues to rapidly grow and expand into a variety of new modalities Peripheral blood has been established as an effective source of autologous progenitor cells Furthermore the graft versus leukemia effect has resulted in novel strategies of adoptive immunotherapy for cancer Finally approaches to gene

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