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Press Release

Tests and studies to diagnose haemorrhages and thromboses

Milan, 19 November 2014 – Diagnostic tests, laboratory analyses and monitoring to assess the risk of haemorrhages and thromboses. And, in the event of necessity, intervention with appropriate treatment. These are the main themes for discussion at the “Congenital and acquired alterations in coagulation” Refresher Course to be held on 19 and 20 November 2014 in Milan by the Fondazione IRCCS Ca’ Granda Ospedale Maggiore Policlinico of Milan and promoted by the Menarini International Foundation. “The Course aims to draw attention to the progress made over recent decades in creating efficient laboratory methods to diagnose the main haemorrhagic and thrombotic diseases”, explains Pier Mannuccio Mannucci, Scientific Director of the Fondazione IRCCS Ca’ Granda Ospedale Maggiore Policlinco of Milan and Chairman of the course.

“The themes range from ischemic events like deep vein thrombosis and pulmonary embolism to events of placentary insufficiency such as the antiphospholipid antibody syndrome. Emphasis is then placed on the evolution of knowledge in the laboratory control of anticoagulant drug therapy, in addition to the criteria adopted to diagnose and monitor treatment of the most frequent congenital coagulopathies: haemophilia, the von Willebrand disorder and platelet function disorders”.

A deficiency in natural anticoagulants is the leading cause of thrombosis. The frequency of antithrombin deficiency in the general population is estimated at around 1 out of 2000/5000 people.
The most frequent symptom is venous thrombosis of the deep veins in the legs, which represents around 90% of all episodes. Thrombosis of the upper extremities, on the other hand, is not a typical symptom of inherited thrombophilia and when it occurs, other causes, often of a local nature, should be looked for. Other thromboses of the cerebral or abdominal veins are more rare (5% of all episodes) but more clinically severe. Superficial thrombophlebitis accounts for the remaining 5% of all symptoms.
Although around half the number of thrombotic events develop apparently spontaneously, for the remaining half, it is necessary to look for and identify contingent contributing causes. Of these, the most important are undoubtedly pregnancy/puerperium and estroprogestinic contraceptives. The onset of thrombotic events during pregnancy/puerperium appears to be more frequent in women with antithrombin deficiency. With these mutations the risk of developing DVT in the leg is 6-10 times higher, compared with women taking the pill with no thrombophilic defects, who in turn have a thrombotic risk 4-6 times higher than women who do not take the pill and have no thrombophilic alterations.

Surgery is another contingent contributing cause to the development of thrombosis in individuals with hereditary thrombophilia. A retrospective study performed in Italy involving a large number of individuals with natural anticoagulant deficiency showed a high frequency of vein thrombosis following abdominal surgery (21%) and high-risk oncological and orthopaedic surgery (37%).

“The purpose of the laboratory investigation is to accurately identify the presence of one or more defects known to be the cause of congenital thrombophilia. Obviously, those individuals with a family history of vein thrombosis must be investigated” continued Mannucci. “Other factors taken into consideration are the patient’s young age at the time of the first thrombotic symptom (under 40-45), recurrence and also its presence at an unusual site (as for example, in the cerebral, mesenteric and portal veins). Thrombotic manifestations during the neonatal period are another indication for study”. For some clinical situations there is little point in carrying out investigations, as for example when venous thrombosis occurs in adulthood or old age, particularly if there is a contingent contributing cause of thrombosis such as surgery, cancer, prolonged immobilization. In these cases, awareness of the existence of a congenital contributing cause of thrombophilia (or lack of one) does not change the therapeutic approach. Moreover, diagnostic tests for thrombophilia are not performed on healthy individuals with no personal or family history of venous thrombosis, when they are electively exposed to contingent thrombotic risk factors as, for

example, pregnancy, high risk orthopaedic surgery and prolonged immobilization.
The course will focus in particular on the von Willebrand (VWD) disorder, which, together with haemophilia A, represents the most frequent congenital defect of haemostasis, with a bleeding tendency of medium severity. Diagnosis is complex but over recent years has become more generally available in our country as in other countries in the world. Treatment too is, generally speaking, excellent.

“Insofar as diagnosis is concerned, a study recently published in the journal Blood (Blood 2014;123(26): 4037-4044) confirms that the BS (Bleeding Score) test is useful not only for confirming the diagnosis, but also for predicting new episodes of spontaneous haemorrhages and, consequently for submitting the patient to pharmacological treatment. The study, conducted in Italy on 796 subjects with various forms of von Willebrand disorder, demonstrated that a bleeding score of over 10 predicts the onset of haemorrhages and the need for treatment”, added Mannucci.

Another study, published in the journal Thrombosis and Haemostasis (ThrombHaemost 2014; 112: 427– 431) showed how the von Willebrand disorder is more severe when the patient in question also suffers from angiodysplasia, a condition that occurs when a small group of tortuous and dilated veins in the mucous of the colon and the small intestine cause bleeding from the lower gastrointestinal tract. Angiodysplasia may occur in one person in four over the age of 60, it is asymptomatic in most people and in most cases remits spontaneously.

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