Dysfibrinogenemias in the Czech Republic

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Introduction: Fibrinogen plays an important role during many physiological processes. Inherited dysfibrinogenemia is a rare disease characterized by functional impairment of fibrinogen molecule. During last decade we have discovered several cases of dysfibrinogenemia in the Czech population.

Materials & Methods: Sequencing of all exons of FGA, FGB and FGG genes as well as fibrin polymerization, fibrinolysis, kinetics of fibrinopeptides release, electron microscopy, coagulation tests and SD-PAGE experiments were performed as described earlier. In some cases the rheological properties of clots were measured.

Results: The molecular cause of congenital dysfibrinogenemia was found in 30 unrelated families in the Czech Republic. In 20 families, mutations were found in the Aα chain. Four mutations were found in the Bβ chain and seven in the γ chain. The most frequent mutations in the Czech population are Aα Arg16His (7 families), Aα Arg16Cys (3 families) and Aα Gly13Glu (5 families). Most patients were asymptomatic following by patients with bleeding tendencies. Thromboses were reported in only four families.

Conclusions: We have investigated about 60 families from the Czech Republic with abnormal coagulation test results suspected with congenital dysfibrinogenemia of which we have found the direct molecular cause of the disease in 30 families. Most patients were diagnosed incidentally (eg prior to surgery) since they did not show any significant clinical manifestation.

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