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Playland.org is the owner and operator of playland.org. This website has no affiliation with IMDb, Sundance Institute, the Hollywood film community, the San Francisco film community, the national film community, the Internet at large, or any other person or entity. All original content is copyright of the individual authors. All other content is copyright of playland.org and may not be used without prior permission. Playland.org is not responsible for the content of external sites. The primary goal of this project is to define the molecular basis of inherited human disorders of hemostasis by identifying the underlying genetic defects. Efforts are currently focused on von Willebrand disease, a bleeding disorder characterized by the absence or abnormal function of von Willebrand factor (VWF). We have used molecular analysis, as well as conventional cloning methods, to identify the genes for VWF and its cofactor, factor VIII, both of which are important in hemostasis. We previously identified a gene, called WVHA, for the X-linked hemochromatosis gene, and we have recently determined the gene structure for this hemochromatosis gene. We also have used the human WVHA gene to develop a transgenic mouse model of hemochromatosis, and this model has been useful in assessing the role of the gene in normal iron metabolism, as well as in evaluating the development of liver fibrosis. We have also identified two genes, called PIK3R1 and PIK3R2, as candidates for the gene for the hyperphosphatasia syndrome, a disorder characterized by excessive bone growth and heterotopic bone formation. We have identified a mutation in PIK3R2 that may be responsible for the molecular defect of this disease. We have also used linkage analysis to localize the gene for beta-thalassemia minor on chromosome 11q, and have identified the beta-globin gene as the candidate gene for the hemoglobin H disease associated with this condition. We have also identified mutations in the gene for factor VII in patients with the Hermansky-Pudlak syndrome, an autosomal recessive disorder characterized by deficient platelet and melanosome formation. We have also studied the genetic defect in the prolactin gene in patients with microprolactinoma, which is the most common pituitary tumor. We have shown that patients with the inherited disorder of vitamin K-dependent coagulation factor II, prothrombin, have a 54b84cb42d

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