Biomarker MIA shows presence of neurofibromas

July 4 2011

Neurofibromatosis (NF1) is a genetic condition which affects one in every 3,000 people. The severity of symptoms can range from benign 'cafe au lait' patches on the skin, through small tumors under the skin and deep plexiform neurofibromas, to malignant tumors of the nerve sheath. New research published in BioMed Central's open access journal *BMC Medicine* shows that a simple blood test for the protein melanoma-inhibitory activity (MIA) could be used to indicate the presence of neurofibromas even if they cannot be seen.

When researchers compared the levels of MIA from blood of patients with NF1 and unaffected controls they discovered that the patients with NF1 had much higher serum levels of MIA and that the level of MIA depended on the number and size of neurofibromas and plexiform neurofibromas the patient had. Tumor biopsies also showed an increase in MIA at the cellular level.

Dr Kolanczyk said, "Using the biomarker MIA to test for the presence and growth of plexiform neurofibromas would be an easier and cheaper way of monitoring clinical course of the patients and would allow the early detection of tumors so improving the treatment, management and outcome. Detection of deep plexiform neurofibroma is currently only possible using MRI scan and since these tumors can become malignant it is important to monitor their growth closely and detect signs of malignant transformation as early as possible."

More information: MIA is a potential biomarker for tumor load in

neurofibromatosis type 1, Mateusz Kolanczyk, Victor Mautner, Nadine Kossler, Rosa Nguyen, Jirko Kühnisch, Tomasz Zemojtel, Aleksander Jamsheer, Eike Wegener, Boris Thurisch, Sigrid Tinschert, Nikola Holtkamp, Su-Jin Park, Patricia Birch, David Kendler, Anja Harder, Stefan Mundlos and Lan Kluwe, *BMC Medicine* (in press)