LETTER TO THE EDITOR

Plexiform mutilating neurofibromatosis – the unpredictable disorder: Whats new?

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We would like to focus the attention in the newly published data which identified two serum markers with potential for early detection of patients with NF1 at risk for developing MPNST, and four markers that could distinguish between patients with NF1 and healthy subjects [1]. Such markers seem to be of extraordinary importance as possible diagnostic tools to support the diagnosis of NF1 and for timely identification of MPNST [1]. The investigations of the colleagues have lead to the identification of four markers (epidermal growth factor receptor, interferon-γ, interleukin-6, and tumor necrosis factor-α), for which significantly different serum concentrations were seen in patients with NF1 compared with healthy controls [1]. Two markers (insulin-like growth factor binding protein 1 (IGFBP1) and regulated upon activation, normal T-cell expressed and secreted (RANTES) showed significantly higher concentrations in patients with NF1 and MPNST compared with patients with NF1 without MPNST [1]. A correlation with internal tumor load was also observed for IGFBP1 [1].

Plexiform neurofibroma, which usually occurs earlier in life, is considered a congenital defect which has a tendency for malignant transformation to malignant peripheral nerve sheath tumors (MPNST) [2, 3]. Malignant progression occurs in 2-16% of patients and could be considered the major reason of morbidity and mortality in NF1 [2, 3]. NF1 seems to be an unpredictable disorder [1]. It varies widely in severity from one person to the next, even between two people in the same family.

No standardized specific treatments for NF1 and its complications are also available [4]. Numerous clinical trials for the treatment of the most common NF1 complications, such as plexiform neurofibromas (Ns) and NF1-related tumors, have been conducted [4]. Unfortunately insufficient safety in in vivo data do not permit the routine use of these medication in the daily clinical practice [4].

References:

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