

## **Abstract**

**The aim of the study** was to evaluate the importance of brain MRI's findings, and modify the criteria for brain MR imaging in NF1 patients according to this data, to improve the quality of life with early detection of important NF1 complications. Description of the whole cohort, with emphasis to possible cause of high range of sporadic NF1 cases. Evaluation the possibility diagnosis or follow up of brain gliomas by plasmatic values of neuron specific enolase (NSE) and S100B protein.

### **Subjects and methods:**

I analysed data from 285 NF1 children followed up on our department from 1990 to 2010 by the same examination battery. I evaluated the incidence of brain MRI findings, clinical development, age at gliomas manifestation and necessity of treatment. I also described the whole cohort and made statistic analysis of plasmatic values of NSE and S100B protein in NF1 patients, with and without brain gliomas.

### **Results:**

OPGs were found in 77/285 (27 %) children and GOOPs in 29/285 (10.2 %) of NF1 children, of who 19 had OPG and GOOP together, so the total number of brain glioma was 87/285 (30.5 %). Totally, 43/87 (49.4 %), respectively 43/285 (15.1 %) children with brain glioma were treated, and 4/285 (1.4 %) of this children died. Obstructive hydrocephalus was found in 22/285 (7.7 %) patients and was caused especially by glioma (14/22) or idiopathic aqueduct stenosis (6/22). Other MRI findings were: hyperintense lesions on T2W images called FASI (Focal Areas of Signal Intensity), cysts, vascular lesions, developmental abnormality, intracranial spreading neurofibroma and perinatal changes on MRI. Parental, especially paternal, age in sporadic cases of NF1 was significant higher than in general population. Plasmatic values of NSE and S100B protein were not significantly different in analysed subgroups.

### **Conclusion:**

All NF1 children benefit from screening brain MRI. We recommend doing the first brain MRI up to the six years of age, because of the highest risk of OPG in this period, and next examinations due to MRI and clinical findings and risk of the other complications. We confirmed an association of advanced parental and particularly paternal age with the occurrence of sporadic NF1. Biomarkers NSE and S100B protein are not suitable for monitoring or diagnosis of gliomas in NF1 children.