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NEUROFIBROMATOSIS VON RECKLINGHAUSEN TYPE 1 (NF1): DIAGNOSTIC CRITERIA AND OTHER IMPORTANT CLINICAL FINDINGS

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Neurofibromatosis von Recklinghausen type 1 (NF1) is autosomal dominant disorder with an incidence of approximately 1:3000-1:4000 individuals. The disease is characterised by the following diagnostic criteria: neurofibromas, café au lait spots, axillary and inguinally freckling, Lisch nodules, optic glioma, distinct osseous lesions, and first degree relative with NF1.
We examined group of 110 children with NF1 (61boys-55%, 49girls-45%). 106(96%) of the patients are alive, and 4(4%) died. All children had MRI examination of the brain. NF1 diagnostic criteria in the examined group were encountered in the following frequency: café au lait spots 109(99%), freckling 102(93%), neurofibroma 93(85%), optic nerve glioma 36(33%), Lisch nodules 61(55%), distinct osseous lesions 9(8%), and first degree relative with NF1 61(55%).
In the group of patients with NF1, further clinical features were found: scoliosis 86(78%), learning disabilities 57(51%), endocrinologic changes 37(34%), brain tumors (without optic nerve glioma) including intraorbital plexiform neurofibroma 29(26%), paravertebral plexiform neurofibroma 22(22%), hydrocephalus 13(12%), epileptic seizures 8(7%), syringomyelia 5(5%), congenital arachnoidal cysts 4(4%) and moya-moya syndrome 1(0.9%). In 92(84%) patients of the NF1 group, one or more high-signal intensity foci were found on the T2 weighted MR images of the brain. Malignancy were found in 7(6%) children.
In conclusion, the knowledge of clinical findings and their frequency is important for clinical diagnosis requires, for long time follow-up, further outpatient and inpatient care.

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