Macrocranium and Macrencephaly in Neurofibromatosis

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Abstract. Data were collected on 52 patients (30 males and 22 females) with neurofibromatosis. Using the volumetric method of Gordon, it was found that 44% of the patients had cranial capacities above the 95th percentile (70% were above the 50th percentile). The presence of intracranial tumors and/or hydrocephalus did not influence skull size. Four patients above the 95th percentile had normal pneumoencephalograms and cerebral angiograms, indicating that the cause of the macrocranium, in some instances at least, is macrencephaly. Volumetric measurement of the sella turcica in 27 of the 52 patients gave results which strongly suggest that idiopathic enlargement of the sella in neurofibromatosis is uncommon, if not rare. Skull films of 26 patients with tuberous sclerosis did not show an increase in cranial capacity similar to that found in neurofibromatosis.

Key words: Neurofibromatosis — Skull abnormality — Sella turcica enlargement — Macrencephaly — Macrocranium.

In 1973, Weichert et al. [12] published the first systematic evaluation of head size in neurofibromatosis. Their study was prompted by the clinical suspicion of macrocranium in two patients with neurofibromatosis from the practice of one pediatrician. They found that of 24 children with von Recklinghausen's disease who had satisfactory roentgenograms of the skull, 75% had macrocranium. As we had not recognized this finding in our practice and had not been able to find it listed in any of the standard texts dealing with neurofibromatosis, we were somewhat skeptical of this high figure but, nevertheless, set out to evaluate our own material. We were aware of the fact during 1972 Norman [6] had reported a family of 10 members with neurofibromatosis, 6 of whom had large heads, and that, in the same year, DeMyer [2], in his excellent paper on megalencephaly in children, had included neurofibromatosis as one of the possible causes. As no references to previous reports of such abnormalities were given, it was assumed that these were isolated, incidental observations.

Case Material and Method of Measurement

In this retrospective study we were able to find in our files 50 patients with neurofibromatosis (28 males and 22 females) who had skull films satisfactory for measurement. These patients ranged in age from 4 months to 62 years with the majority being in the pediatric age group. Cranial capacity was calculated from 68 skull examinations obtained on these patients, using the well known method of Gordon [4]. This approach, which measures the inner capacity of the skull, is in contrast to the study by Weichert, et al. [12] in which outer size of the skull was measured by the method of Haas [5]. Gordon’s method was chosen because we wanted to get some idea of cranial content size and because it embodies percentiles rather than the statistically less acceptable “range” inherent in the Haas procedure. Furthermore, it seemed reasonable to assume that confirmation of macrocephaaly in neurofibromatosis by a different mode would be even more convincing. Incidentally, Bray et al. [1] have reported a close correla-
tion between Gordon's figures and the widely used clinical measurement of occipito-frontal circumference.

Patients with known intracranial tumors and/or hydrocephalus were included in the initial study.

Results

The results are shown in Figure 1. As can be seen, 80% of the boys' skulls and 75% of the Girls' skulls had cranial capacities greater than the 50th percentile. Of greater significance is the figure obtained for the abnormally large heads. No less than 46% of the 50 patients had cranial capacities above the 95th percentile.

Later, when our total number of patients had grown to 52, we used a somewhat more uniform approach, selecting only the initial skull examination for each patient. In this analysis the total number of patients with cranial capacities above the 50th percentile dropped to 70% but the more important group, those patients with cranial capacities above the 95th percentile, remained almost constant at 44%.

We again chose to include those neurofibromatosis patients with known intracranial tumors and/or hydrocephalus in our analysis. In this group there were six optic nerve gliomas, three pontine gliomas, two patients with aqueduct stenosis and one each with acoustic neuroma, cerebellar astrocytoma and communicating hydrocephalus. One older patient, originally thought to have hydrocephalus, was subsequently shown to have cerebral atrophy. Although these patients with known intracranial lesions were included, they were identified in such a way on the graphs that one could determine if their presence in any way influenced the overall results as far as cranial capacity was concerned. That they did not is clearly shown in Figure 2. It is evident that in patients with known intracranial pathology, 70% are above the 50th percentile and 44% are above the 95th percentile, indicating no difference whatsoever from the previous determinations.

Among the neurofibromatosis patients who had special neuroradiologic procedures such as angiography and pneumoencephalography, there were four who had cranial capacities above the 95th percentile.