Among the tumours and other space-occupying lesions of the central nervous system there are some congenital conditions with obvious pathological birthmarks on the skin, retina and other organs. These lesions, which have been studied pathologically and clinically during the last 80 years, perhaps can be grouped under the general term of phakomatoses of van der Hoeve (phakos = birthmark). No doubt some pathologists would prefer other denominations, like hamartomas or hamartoblastomas because they consider the term phakomatoses inadequate. However, they do agree about the overlapping of the different diseases included in this large group with cutaneous manifestations and involvement of peripheral and central nervous systems.

An interesting feature of many of these conditions is that they appear as borderline cases between congenital malformations and tumours. Norman considered that some of these anomalies belong to the category of hamartomas or congenital malformations with a potentiality for growth. The general term of hamartoblastomatosis was preferred by Zülch to the one of phakomatoses and this author also referred to the classical concept of Bielschowsky considering these lesions as developmental disorders with a tendency to blastomatosis (“Entwicklungsstörungen mit blastomatosem Einschlag”).

Material

In our series of 2,200 brain tumours and other expansive intracranial lesions and 275 cases of different types of spinal-cord compression that we have operated on in Madrid during the last 16 years, we have had several examples of the so-called phakomatoses.

We shall present very briefly the main features of 12 patients with very obvious cutaneous and peripheral abnormal birthmarks together with other neurological symptoms who have been submitted to neurosurgical procedures.

Although there is some confusion in the literature about the different types and the numbers of the so-called phakomatoses we shall consider only three main groups of practical and neurosurgical interest: (i) neurofibromatosis (von Recklinghausen’s disease); (ii) tuberose sclerosis (Bourneville’s disease); and (iii) the wide group of angiomatosis which includes the encephalofacial angiomatosis of Sturge-Weber, the retinocerebellar angiomatosis of von Hippel-Lindau and other more rare types of cutaneous and nervous angiomatosis.

Neurofibromatosis
(von Recklinghausen’s Disease)

The clinical and pathological manifestations of von Recklinghausen’s neurofibromatosis are well known. Besides the typical cutaneous pigmentations and the multiple peripheral and subcutaneous tumours, there may be associated malformations of the central nervous system. But more interesting from our point of view is the occasional presence in these patients of intracranial or spinal tumours. Neurofibromas, meningiomas and different types of gliomas have been described.

Two of our patients with von Reckling-
hausen’s disease (19 and 32 years old) presented signs of spinal-cord compression and at the operations, carried out by our co-worker Dr. Boixados, multiple intradural neurofibromas were removed from the cervical region in 1 and from the dorsal region in the other. Postoperative results were good in both cases, but 1 patient presented a recurrence of symptoms 4 years later and another large neurofibroma was removed from the same cervical region.

In 2 other patients (28 and 35 years old) with the typical manifestations of von Recklinghausen’s disease and verified peripheral neurofibromas there appeared symptoms of an increase of intracranial pressure. A frontal meningioma was removed in 1 of these patients and in the other only a decompression was performed after the air studies showed a basal tumour in the region of the 3rd ventricle.

The association of von Recklinghausen’s disease with gliomas of the optic nerves and chiasm also was observed in 2 of our patients with histologically verified optic gliomas and the typical cutaneous pigmentations.

But our most interesting example of this group concerns a case of glioma of the 4th ventricle which already has been reported in detail. A woman 45 years of age had a clear von Recklinghausen’s disease of hereditary basis, as the cutaneous and peripheral manifestations (Fig. 1) also were very marked in another sister 30 years old. The biopsy of one of the subcutaneous tumours of the patient demonstrated a typical neurofibroma. Besides there were symptoms of vertigo, headaches, vomiting and difficulty in walking and standing. Papilloedema, nystagmus, tremor and profound disturbances in the gait were the most outstanding signs. The possibility of acoustic tumours was disregarded as the result of otological examinations, and ventriculography revealed a filling defect in the 4th ventricle, besides a marked dilatation of the lateral and 3rd ventricles.

At operation a reddish, soft tumour filling the 4th ventricle and implanted in its floor was removed. The histological sections (Dr. Escalona) demonstrated an astrocytoma of fibrous type, with marked hyperplasia of the neuroglia fibers (Fig. 2).

The evolution of this case has been quite good during the year passed after the operation and she is greatly improved.

From the histological point of view there may be doubts and discussion about the classification of some of the central lesions that appear in the neurofibromatosis of von Recklinghausen. The so-called “central neumomas” of the old literature may be considered as malformations by some pathologists or under different names of tumours by others (astrocytomas, spongioblastoma polare, schwannoma, etc.). As we have emphasized before, the lack of sharp boundaries of such lesions confers a special interest in this group and in the following group in which malformations, anomalies of development and growth of tumours sometimes are closely related within the central nervous system of those dysplastic and dysontogenetic organisms.

**Tuberose Sclerosis**
(Bourneville’s Disease)

There is no need to recall the great diversity of lesions in this condition affecting the skin, brain and other organs, such as kidneys,