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Hybrid Phakomatosis: From Initial CT Observation to Molecular Studies

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Vertebroplasty and Kyphoplasty: Is One of These Procedures the Best Choice for All Patients?

Vertebral compression fractures (VCFs) are a widespread problem in elderly populations, with approximately 700,000 VCFs occurring in the United States each year as a result of osteoporosis. Vertebroplasty and kyphoplasty are two minimally invasive procedures that are increasingly used to treat VCFs. Both procedures stabilize VCFs by injecting bone cement into the vertebral body, with kyphoplasty having the additional step of reducing the fracture with an inflatable bone tamp (IBT).

Both of these procedures provide excellent pain relief for most patients, with a low incidence of serious complications. It has been suggested, however, that kyphoplasty may afford a lower risk of clinical complications than vertebroplasty. Use of the IBT can elevate the vertebral endplates toward their pre-fracture levels and create an intravertebral cavity. This cavity allows for the injection of more viscous cement under lower pressure, which may result in a lower rate of clinically significant cement leaks.

In addition to creating a cavity, the potential fracture reduction would restore vertebral height and correct kyphotic deformity. This can reduce the risk of serious comorbidities related to kyphotic deformity, many of which involve pulmonary dysfunction. It has also been suggested that kyphotic deformity resulting from VCFs might increase the risk of subsequent VCFs (1).

Recently, there have been reports of height restoration and kyphosis correction in a few vertebroplasty studies. These studies found the greatest degree of height restoration in vertebrae exhibiting intravertebral clefts, or pseudarthrosis. In some of these studies, physicians were able to manipulate the spine through padding and positioning during the procedure. Although height restoration and kyphosis correction were observed, they were not to the same degree as that observed in published kyphoplasty studies (2).

The low cost and short procedural time associated with vertebroplasty make the procedure an attractive choice for VCFs not exhibiting a high degree of kyphotic deformity or vertebral height loss. Although vertebroplasty may successfully treat the pain experienced by patients with severe vertebral deformity, it does not address deformity to the same degree as kyphoplasty. Because of the risks and comorbidities often associated with kyphotic deformity, it is a common symptom of VCFs that needs to be addressed.

The degree of height restoration and kyphotic deformity correction experienced by kyphoplasty patients leads me to believe that it may become the standard of care for VCFs under certain circumstances. In a 2003 publication by Fourney and colleagues (3), both procedures were performed using a decision tree to determine which procedure was best for which patients. They performed kyphoplasty for patients with significant vertebral collapse, kyphosis greater than 20°, or disruption of the posterior vertebral cortex, whereby more controlled cement delivery was advantageous to minimize risks of spinal compromise. In the future, I believe many clinicians may use decision trees of this nature to determine which procedure is best for each individual case.

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Hybrid Phakomatosis: From Initial CT Observation to Molecular Studies

We read with much interest the case report by Erbay et al (1) dealing with rapid development of optic glioma in an infant with both neurofibromatosis type 1 (NF-1) and tuberous sclerosis (TS).

The neoplasm showed an aggressive behavior that is not commonly seen in children with NF-1, and the authors hypothesized that it might be due to what they called "hybrid" phakomatosis (ie, the coexistence of more than one phakomatosis). They concisely but accurately illustrated growing evidence in the literature indicating the genetic basis for TS- and NF-1-induced neoplasms. The formation of neoplasms in both conditions appears to be due to deficiency of tumor-suppressor genes. Although the TS and NF-1 genes are different, they appear to have common enzymatic pathways at the cellular level. Therefore, there might be an additive or synergistic effect in patients with both of these disorders.

Coexistence of more than one phakomatosis is a rare condition, and Erbay et al have been able to find only eight cases reported in the literature. Their article, however, reminded us of what we observed some time ago, in the late 1970s, when the phakomatoses were a still "mysterious" part of pathology and their pathogenesis was poorly understood. In a series of 65 cases of phakomatoses examined with CT, we found three patients who simultaneously had features of two different phakomatoses. Two of the three, reported in 1980 (2), had the typical clinical features of NF and also had subependymal or intranuclear nodular calcifications showing a CT appearance identical to that of tubers in TS. At that time, CT had just become the criterion standard for neuroimaging, and it was emerging as the most valuable method to investigate patients with phakomatosis (3). It provided both an early diagnosis, by displaying all orbital and craniocerebral lesions, and a suitable follow-up of these patients, who appeared prone to develop orbital and cerebral tumors.

Time has passed, and there have been advances in the diagnostic criteria of the phakomatoses. MR imaging is now widely used for the characterization of brain lesions associated with these disorders, and their molecular genetic aspects are being understood. Thus, Erbay et al could possibly explain the abnormal predisposition to neoplasm formation that we too found in what we defined as "atypical" and they as "hybrid" phakomatosis. As a matter of fact, either of the patients with both NF and TS showed an abnormal predisposition to neoplasm formation. The 27-year-old man with café-au-lait spots in case 1 had undergone surgery a few years earlier for a frontal meningioma (a rather unusual tumor in men and in young people, in general), and his CT scan showed a cerebellar tumor in addition to bilateral acoustic neuromas. The 12-year-old girl with cutaneous manifestations of NF in case 2 presented with left hemiparesis and was found to harbor both a right deep temporal glioma and neoplastic infiltration of the left optic

nerve, optic chiasm, and right optic tract. The significant advances in the genetics of these disorders made in the past few years may help us understand why.

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