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Phacomatoses in the pediatric age group

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Abstract
The most common phacomatoses in children that need surgical attention are neurofibromatosis 1 and 2, tuberous sclerosis complex, Sturge-Weber disease, Von Hippel-Lindau disease, and neurocutaneous melanocytosis. All are rare and, as genetically determined disorders, all complex multisystem diseases with multiple manifestations outside the CNS. Diagnostics, management recommendations, and surgical care are age-specific and require individualization. The lifelong multidimensional disease burden demands a multidisciplinary and well-coordinated management approach. The consequence of these boundary conditions is that management of children with a phacomatosis is everything else but simple, straight forward, and intuitive. This Special Annual Issue is designed to serve as an up-to-date encyclopedic reference for all aspects of management of phacomatoses in the pediatric age group.

Keywords Phacomatoses · Pediatric age group · CNS · Neurofibromatosis type 1 · Neurofibromatosis type 2 · Tuberous sclerosis complex · Sturge-Weber disease · Von Hippel-Lindau disease · Neurocutaneous melanocytosis

Preface

Phacomatoses are rare genetic syndromes, thus seen quite seldom in general practice, and tend to concentrate in dedicated centers for specific disease types. As multisystem diseases, only a portion of their symptoms and pathologies manifests within the CNS, or within the treatment scope of pediatric neurosurgeons. Note that some aspects, even though manifested within the CNS, cannot be treated surgically. Furthermore, phacomatoses encompass a diverse kaleidoscope of phenotypes, resulting in very individual disease constellations. Thus, knowing or having identified the underlying genetic disorder is far from being a blueprint for further disease management. Furthermore, management in the pediatric age group is more complex than that in adults. Manifestations and complications in infants, toddlers, children, and adolescents (younger and older) are very age-specific in diagnostics and care, in terms of patient needs and demands, compliance, and other limitations. The initial consultation might, in some cases, begin with prenatal counseling when one of the parents harbors the disease, or when an unexpected intrauterine finding emerges.

Therefore, an interdisciplinary team comprising several pediatric subspecialities, different surgical disciplines apart from pediatric neurosurgery, dedicated pediatric imaging teams, human genetics, social workers, and psychologists, is necessary to provide comprehensive treatment, not only to the affected child but also to possibly affected family members.

Focusing on the pediatric neurosurgical treatment aspect, the situational complexity is similar. Limited transferability exists for "rules" regarding operative treatment for similar manifestations, such as tumors, from the situation in children without a phacomatosis background. Most of the time, the simple existence of a tumorous lesion does not define the indication for surgery. On the contrary, thorough knowledge of the expected natural course, the recent tumor dynamics, the results of the complete diagnostic work-up (*where do we stand?*), and the co-existence of other problematic disease-specific manifestations (epilepsy, extra CNS manifestations, etc.) is required to define the surgical

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intention correctly and to determine the ideal time point for a surgical intervention. Often, a decision to *not* operate is wiser than immediate surgical intervention. On the other hand, in situations such as devastating epilepsy, surgical intervention must not be delayed for too long.

Since the diagnosis of a phacomatosis means a lifelong burden for the affected children, which may lead to several surgical interventions for an individual over the course of a lifetime, care must be taken to expose the child to only the *minimal* amount of effective (and not harmful) surgery necessary to serve the child's needs and provide benefit in the best possible way.

The transition from adolescence into the adult world must be mediated, as the patients grow into a world where their problems do not lessen, yet their supportive structures are often less developed. Especially for those young patients above 20 years with cognitive deficits, life as a self-caring "adult" is impossible.

It was our great pleasure and objective to create a comprehensive update on the major types of phacomatoses that pediatric neurosurgeons would encounter, like NF1, NF2, TSC, VHL, SW, and NCM. We have tried to include all significant non-surgical perspectives, as well as sharing surgical insights from disease-specific surgical expert teams. We are most grateful to all the author groups, especially the "non pediatric neurosurgeons", many from renowned institutions, who have sacrificed precious time during the currently ongoing, challenging SARS-CoV-2 pandemic, to create this Special Annual Issue, designed as an up-to-date encyclopedic reference for the management of phacomatoses in the pediatric age group.

The willingness of all participants to contribute and to help educate the worldwide pediatric neurosurgery community, as well as their discipline to "deliver in time," was amazing and selfless, demonstrating that we are all part of a greater enterprise to improve the lives of sick and affected children everywhere.

Compliance with ethical standards

Conflict of interest SC and MUS have no conflict of interest to report.