Focal areas of high signal intensity (FASI)

Neurofibromatosis type 1 (NF1) is the most common neurocutaneous syndrome, affecting 1 every 2500–3000 individuals. In 90% of cases it arises from an autosomal dominant mode of inheritance from mutation of the tumour suppressor NF1 gene located at chromosomes 17q11.2. In the other half of cases it can arise from a de-novo mutation.

The disease has a multi-systemic manifestation affecting the orbits, brain, spine, musculoskeletal system, breast, lung and skin. Many patients have a tendency to present in childhood, however, although uncommon, it has also been known to present in adulthood.

CNS manifestations of NF1 can be roughly divided into three main areas where pathology is seen, which are the intracranial, orbital and spinal regions.

NF1 can have serious consequences for young patients presented with such a diagnosis. The most common neurological disability seen in children is cognitive difficulty, which is almost always irreversible and carries through to adulthood.

NF1 has also been associated with other significantly debilitating neurological conditions including epilepsy and multiple sclerosis (MS).

In this presentation we provide a pictorial review of patients that we have encountered at our regional specialist children’s centre presenting with neuroimaging manifestations of NF1, and provide overviews of the relevant features of each.

Radiological Findings

Lateral Thoracic Meningoceles

Lateral thoracic meningeoceles are defined as the herniation of the thecal sac through the spinal foramen (arrow).

Over time the neural foramen is eroded and the protrusion of thecal sac causes development a dumbbell-shaped appearance that, akin to meningioma's, is mimicked on T1/T2 with surrounding cerebrospinal fluid signal intensity.

Plexiform Neurofibromas

Plexiform neurofibromas are an uncommon variant of neurofibromas that involve long nerve segments and their associated branches.

Although they are generally benign WHO grade I tumours, there is a significant potential for malignant transformation, which may occur in 5-10% of larger lesions.

On T1W MRI, they appear hypointense while on T2W imaging they are seen as herring a hypointense rim (red arrow), due to CSF with a hypointense central focus (blue arrow) due to collagenous stroma. These features together give a typical 'target sign' appearance.

Sphenoid Wing Dysplasia

Sphenoid wing dysplasia is a characteristic, but not a pathognomonic feature of NF1, it is defined as one of the diagnostic criteria for the condition.

The greater wing of sphenoid specifically is generally the more commonly affected site.

Dysplasia of the sphenoid wing is often associated with plexiform neurofibromas and together, may cause herniation of temporal lobes into the orbit.

Sphenoid Wing Dysplasia

Numerous cavernous malformations may develop as a result of the ischaemia. In the earlier angiography/CT angiography with 3D reformats (blue arrows) this is seen as a characteristic puff of smoke' appearance, literally translated in Latin to 'moyamoya'.

Development of arteriovenous malformations, vascular ectasia and cerebral aneurysms may also be seen.

Conclusion

NF1 is the most common phakomatosis, affecting multiple sites and organ systems. CNS manifestations occur in roughly 10-20% of patients and include regions of myelin vacuolization (FASIs); multiple tumours including optic nerve gliomas, cerebellar astrocytomas and optic nerve-hypoplasia, vascular abnormalities, and dural ectasia. Understanding the characteristic imaging findings, as well as their clinical significance and expected evolution, is essential when interpreting neuroimaging studies in this patient population.