NEURORADIOLOGY

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NEUROFIBROMATOSIS TYPE 1 (NF1) IN CHILDREN AND UNIDENTIFIED BRIGHT OBJECTS (UBOS) ON MAGNETIC RESONANCE IMAGING (MRI).

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Introduction: MRI of the brain in children with NF1 may show areas of increased T2 – weighted signal intensity called UBOs present in 43% up to 92% of the cases. As UBOs are almost constantly observed at an age in which all signs of the disease are not present, they are of great interest for the diagnosis.

Objetives: To investigate the frequency and location of UBOs in patients with NF1.

Materials and methods: Observational retrospective study. Brain MRIs of 83 patients aged 0 to 21 years were included. 43 suffered from NF1 and 40 had a MRI performed at the hospital for any other condition unrelated to NF1.

Results: UBOs were identified in 53% of patients with NF1and in 67% of the children between 4-12 years old. No patient aged 0-3 years nor controls presented UBOs**.**Cerebellum and globuspallidum were the predominant affected areas (35% and 33% respectively) The sensitivity of the presence of UBOs was 53% with a false negative rate of 47%,a specificity of 100% and a false positive rate of 0%.

Conclutions: When diagnostic criteria for NF1 are not fully present -especially in children aged 4 to 12 years- high frequency and specificity of UBOS identified by MRI can be used as a complementary diagnostic tool.

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INHERITED MANGANISM: TYPICAL CLINICAL AND NEUROIMAGING FEATURES

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INTRODUCTION: Inherited Manganism (IMn) is an inborn error of manganese (Mn) homeostasis. This unusuall entity is comprised by polycythaemia, movement disorders, liver cirrhosis, hypermanganesaemia, and peculiar hyperintense signal in the basal ganglia in the T1-weighted sequence of MRI.

CASE DESCRIPTION: An 11-year-old girl with uneventful development until the age of 7 years, when presented progressive cognitive impairment and limb dystonia. Furthermore, she had polycythemia, chronic liver disease and high levels of serum manganese. MRI disclosed bilateral hyperintense signal in the basal ganglia, midbrain and cerebellar nuclei in the T1-weighted sequence, suggesting Mn storage (figure). It was detected mutations in SLC30A10, accountable for a manganese transporter.

DISCUSSION: IMn is an inborn error caused by mutations in the Mn transporter gene SLC30A10. It's characterized by hepatic cirrhosis, polycythemia, hypermanganesaemia, and a severe syndrome extrapyramidal. Typical brain MRI in IMn comprises: hyperintense signal in the globuspallidus on T1-weighted sequences, with changes extending into adjacent basal ganglia in most cases. White matter involvement may also be observed. T2 changes of low signal return from the globuspallidus are also present. Treatment includes chelating therapy and iron supplementation.

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AN UNUSUAL CAUSE OF BLINDNESS: BILATERAL GENICULATE LESION.

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Introduction: The geniculate bodies are a particular region topographically of central nervous system which are vascular the anterior and posterior choroidal artery. By this characteristic are rare vascular lesions that lead to an injury in this region. We report a case of a child who presents with this peculiar injury after a frame of diarrhea.

Case description: A 10 years girl presented with acute blindness one day after a moderate episode of febrile diarrhea. Examination showed no signs of dehydration, normal pupillary response to light but complete bilateral amaurosis. Supplementary exams included electrolytes, were normal. MRI revealed within the lateral geniculate bodies areas of symmetric bilateral decreased signal intensity on T1-weighted imaging with correspondent hyper-signal-intensity on T2-weighted images. These areas were consistent with acute infarcts that became slightly hemorrhagic.

Conclusion: Due the lateral geniculate bodies be in watershed topography, they are susceptible to injury by hypoperfusion leading to an infarct before the effectiveness of the anastomotic network.

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CLINICAL SPECTRUM AND NEUROIMAGING IN CHILDREN WITH MALFORMATIONS OF CORTICAL DEVELOPMENT

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Introduction: Malformation of cortical development (MCD) includes wide range of neurodevelopmental disorders.

Aim of work: to present the clinical spectrum of MCD in relation their identified types, EEG & neuroimaging data.

Subject & methods: forty five case with MRI findings of MCD, aged from 14 days -4 years ,were subjected to clinical evaluation ,EMG & NC, ABR , echocardiography ,abdominal US,karyotyping,TORCH screening, TMS/MS, lactate level (7 cases each) , urine GAG and CPK(one case each)

Results: lissencephaly / pacchygyria spectrum comprised 57.8% of cases schizencephaly (17.8%), polymicrogyria (13.3%), tuberous sclerosis (TSC) (6.7%), hemimegalencephaly & holoprosencephaly (2.2% each). Microcephaly was present in (77.7%),seizures occurred in (24.4%) :infantile spasm 2 cases (4.4%), multiple seizure types 7 cases (15.5%), myoclonic seizures (4.4%) and they were refractory in 7 cases .Suggested etiologies were ;TSC in 3 cases(6.7%), KlippleTrenauny , muscle eye brain disease (MEBD) ,CMV infection and prenatal insult one case each

Conclusion: MCD should be considered among patients with developmental delay, microcephaly and seizures. Lissencephaly was the commonest followed by schizencephaly Microcephaly& refractory seizures were more significant among cases with lissencephaly / pachygyria compared to other types of MCD. Aetiological diagnosis such as TSC, MEBD, TORCH infection are important for management. Molecular diagnosis of MCD is necessary, hence prenatal diagnosis

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THE SPECTRUM OF LEUKODYSTROPHIES IN CHILDREN: EXPERIENCE AT A TERTIARY CARE CENTRE FROM NORTH INDIA

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Introduction: The term 'leukodystrophies' refers to the disorders with primary white matter involvement with demonstrable biochemical or molecular defect and usually with a progressive clinical course. This study aimed to describe the spectrum of leukodystrophies managed at a tertiary care and referral centre in north India.

Methods: The medical records of children diagnosed with a leukodystrophy at a tertiary care, referral hospital in North India from January 2008 to December 2012, were retrospectively reviewed. The diagnosis was based on the clinical phenotype, suggestive neuroimaging and definitive investigations where applicable and available. The data was extracted as per a pre-designed proforma. The clinical and radiological data of each case was subsequently summarized and reported.

Results: During the study period, 83 cases were diagnosed as a leukodystrophy. The white matter disorders with demyelination (27/83; 32.5%) were the most common (Metachromatic leukodystrophy-12; X-linked Adrenoleukodystrophy-8; Krabbe Disease-5; Alexander Disease-2). This was followed by hypomyelinating disorders (26/83; 31.3%; Pelizaeus-Merzbacher Disease-10; Pelizaeus-Merzbacher like

disorders-1; GM1 gangliosidosis-2; GM2 gangliosidosis-2; Fucosidosis-1; Cockayne Symdrome-1; 4H syndrome-1; Unknown etiology-8). The other disorders included Megalencephalicleukoencephalopathy with subcortical cysts (21), Canavan Disease (3), Vanishing white matter disease (2), Suspected Aicardi-Goutieres syndrome (1), Leukoencephalopathy with brainstem and spinal cord involvement and lactate elevation (1) and Leukoencephalopathy with anterior temporal lobe cysts (2).

Conclusions: The clinical and radiological clues may be helpful in guiding the investigations of a child with suspected leukodystrophy.

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PHELAN-MCDERMID SYNDROME ASSOCIATED WITH POLYMICROGYRIA

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The 22q13.3 deletion causes a neurodevelopmental syndrome, also known as Phelan-McDermid syndrome (MIM #606232), characterized by developmental delay and severe delay or absence of expressive speech. We describe a 2-year-old girl with severe developmental delay carry a 22q13 terminal deletion associated to ring 22 chromosome who had left opercularpolymicrogyria associated with right retrocerebellar cyst. Polymicrogyria (PMG) is a malformation of cortical development due to an abnormal organisation. It is a heterogeneous disorder associated with genetic and acquired events, namely 22q11.2 deletion syndrome also known as DiGeorge syndrome (DGS) /velocardiofacial syndrome (VCFS) among others. We suggest that in children with 22q alteration/deletion, polymicrogyria should be ruled out.

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ARTERIAL SPIN LABELING PERFUSION MRI ANALYSIS IN A PATIENT WITH BRAIN SWELLING FOLLOWING ACUTE SUBDURAL HEMORRHAGE

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The brain swelling following acute subdural hemorrhage, so-called 'bigblack brain' has been reported in infants, while the pathophysiology remains unclear so far. We report a fourteen-month-old boy who showed brain swelling four days after acute subdural hemorrhage and performed arterial spin labelling (ASL) perfusion MRI analysis. The patient was delivered to our hospital because of right hemiconvulsion and decreased mental status after falling down from the table and head banging. He showed right hemiparesis. Brain CT showed acute subdural hematoma on the left hemisphere and mild midline shift without a sign of loss of gray-white matter differentiation. On the same day, hematoma evacuation was operated. Although the hemiparesis was gradually improving in a few days, repetitive vomiting and left-sided eye deviation appeared three days after head trauma and mental status and right hemiparesis were aggravated again. Brain CT showed loss of gray-white matter differentiation on the left hemisphere. Brain MRI on day 5 demonstrated restricted diffusivity in left cerebral white matter. ASL perfusion MRI showed left cerebral hyperperfusion. One month after traumatic brain injury, he had mild right hemiplegia, right facial palsy and left hemianopsia. Brain MRI showed left cerebral atrophy and disappearance of hyperperfusion in the left hemisphere. In recent years, the trigeminovascular system, which is neurogenic regulation system of cerebral vessels, is possible to explain the 'bigblack brain' pathophysiology. In conclusion, MRI analysis in our case supports the idea that trigeminovascular system causes brain swelling following acute subduralhemorrhage.

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COMPARISON OF CRANIAL MAGNETIC RESONANCE IMAGING FINDINGS AND CLINICAL FEATURES IN PATIENTS WITH CORPUS CALLOSUM ABNORMALITIES

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Aim: To evaluate the relationship between clinical and cranial magnetic resonance imaging findings in patients with corpus callosum abnormalities.

Materials and methods: Between September 2010 - March 2012, patients with developmental corpus callosum abnormalities were included in the study. Corpus callosum abnormalities were classified as total agenesis, partial agenesis and callosal hypoplasia. Regarding the groups, the association between radiological abnormalities and clinical findings were evaluated.

Results: A total of 62 patients (32 female (51.6%) and 30 (48.4%) male) with a mean age of 18.0±32.1 months were enrolled in the study. Twenty patients (32.3%) had total agenesis, 9 patients (14.5%) had partial agenesis and 33 (53.2%) patients had hypoplasia of the corpus callosum. Thirty five (56.7%) cases had abnormal physical examination, 47 (75.8%) cases had abnormal neurological examination and 42(67.7%) cases had psychomotor retardation. There were not significant differences among groups regarding physical examination, psychomotor retardation, seizures or microcephaly. Seizures, psychomotor retardation and neurological abnormalities were significantly more frequent in patients with associated other radiological abnormalities. Posterior segment of the corpus callosum was significantly thinner in patients with psychomotor retardation and the anterior part of the corpus callosum thinner in patients with abnormal physical examination. Patients with total agenesis were more prone to seizures at an early age than patients with partial agenesis or hypoplasia.

Conclusion: The neurologic prognosis of patients with corpus callosum abnormalities is poorer in patients with an associated neuroradiological abnormality. Early development of seizures may be observed in cases with total agenesis of corpus callosum.

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GENERALISED RADIOLOGICAL ABNORMALITY ASSOCIATED WITH ACUTE NEUROLOGICAL PRESENTATIONS IN SICKLE CELL DISEASE

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Introduction: Sickle cell disease (SCD) is the commonest cause of stroke in childhood. Ninety percent of patients have radiological or pathological evidence of large vessel disease. However, patients presenting with neurological symptoms and signs after chest crisis have been reported to have generalised, rather than focal, neuroradiological abnormalities (posterior leukencephalopathy and acute demyelination). As few cases have been reported, the pathophysiology and natural history remain obscure.

Methods: As part of a 10-year prospectively collected registry of children with SCD, we report our experience with patients presenting acutely and found to have generalised radiological abnormality.

Results: Of 51 patients documented to have had an acute neurological presentation with focal signs, seizures or coma, 4 (8%) had generalised rather than focal neuroradiological abnormality on imaging within 3 days of presentation. 3 had generalised cerebral oedema, of whom 2 had bilateral borderzone infarction involving grey as well as white matter. These patients survived and reintegrated into mainstream school without significant motor disability; none has had a recurrence after follow-up of 4-10 years. The fourth child had posterior leukencephalopathy radiologically; he recovered consciousness but died of his pre-existing renal disease.

Conculsion: Our patients extend the neuroradiology associated with acute seizures and coma in SCD to include generalised cerebral oedema and bilateral borderzone infarction as well as posterior leukencephalopathy. The neurological outcome may be favourable if the patient survives the acute phase. The role of acute hypoxia and blood pressure abnormalities requires investigation.

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N-METHYL D-ASPARTATE RECEPTOR (NMDAR) ANTIBODIES ASSOCIATED WITH DISTINCT CLINICO-RADIOLOGICAL WHITE MATTER SYNDROMES: CLINICAL EVIDENCE FOR AN ANTI-NMDAR LEUKOENCEPHALOPATHY?

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Introduction: NMDAR-Ab encephalitis is characterized by seizures, movement disorder and psychiatric symptoms. Cases comprising of a predominant white matter disorder have recently been reported to be associated with NMDAR-Ab.

Method: Ten children with significant white matter involvement, with or without NMDAR-Ab encephalitis, were identified from 46 consecutive NMDAR-Ab positive pediatric patients. Clinical and neuroimaging features were reviewed and the treatment and outcomes of the neurological syndromes evaluated.

Results: Three distinct clinico-radiological phenotypes were recognized: brainstem encephalitis (n=3), leukoencephalopathy following herpes simplex virus encephalitis (HSVE; n=2) and acquired demyelination syndromes (ADS; n=5). Three of the ADS had myelin oligodendrocyte glycoprotein (MOG) as well as NMDAR-Abs. Typical NMDAR-Ab encephalitis was seen in 3 patients remote from the first neurological syndrome (2 brainstem, 1 post HSVE). Six of the seven patients (85%) who were treated acutely, during the original presentation with white matter involvement, improved following immunotherapy with steroids, intravenous immunoglobulins and plasma exchange, either individually or in combination. Two patients had escalation of immunotherapy at relapse resulting in clinical improvement. The time course of clinical features, treatments and recoveries correlated broadly with available serum antibody titers.

Conclusion: Around 20% of children with NMDAR-Ab have clinico-radiological evidence of white matter involvement, often distinct from NMDAR-Ab encephalitis, and appear immunotherapy responsive, particularly when treated in the acute phase of neurological presentation. When observed, this clinical improvement is often mirrored by reduction in NMDAR-Ab levels, suggesting that these antibodies may mediate the white matter disease.