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RARE CAUSES OF EPILEPSY IN PAEDIATRIC NEUROIMAGING A CROSS SECTIONAL PICTORIAL ESSAY

Radiology

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ABSTRACT

Paediatric neuroimaging is a super speciality branch of radiodiagnosis. Majority of patients present with seizures. These cases would aid the general radiologists in diagnosis of such rare conditions .We would like to share a few cases we encountered during our clinical practise . We present it as a pictorial essay as radiology is a visual science providing aids for easy diagnosis for our fellow colleagues . We present a pictorial assay of five rare disorders which we encountered in our practise over a period of three years. We would be presenting the imaging findings followed by a brief discussion about each condition .

KEYWORDS

PVL, Hypomelanosis of Ito, Dyke –Davidoff-Masson syndrome , Rasmussens encephalitis , Crouzons syndrome.

INTRODUCTION:

Magnetic resonance imaging is widely used to evaluate children having seizures. We would discuss five very rare pathologies encountered in paediatric neuroimaging . Each of these pathologies have very characteristic findings which aid their diagnosis. We have included severe periventricular leukomalacia which is a form of end stage of hypoxia induced encephalopathy , Hypomelanosis of Ito a rare phakomatosis , Dyke –Davidon Masson syndrome a rare congenital anomaly , Rasmussens encephalitis an end stage disease of previous encephalitis and Crouzons syndrome a very rare craniofacial dysostosis thus including post hypoxic, post infective and genetic abnormalities.The imaging findings of these conditions are absolutely diagnostic hence aid in their diagnosis and prevent unnecessary further work up .

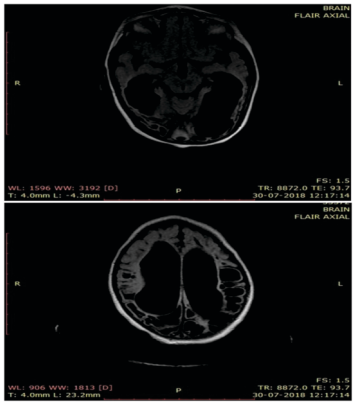
TECHNIQUES AND METHODS :

Magnestic resonance imaging ,FLAIR sequences ,T2WI and FLAIR axial, FLAIR coronal , T1WI post contrast imaging in axial sections and CT of Brain bone windows. In this series we would also emphasize the accuracy of dignostic features aiding to prompt diagnosis of these conditions hence establishing the sensitivity of Magnetic resonance imaging .

OBSERVATION:

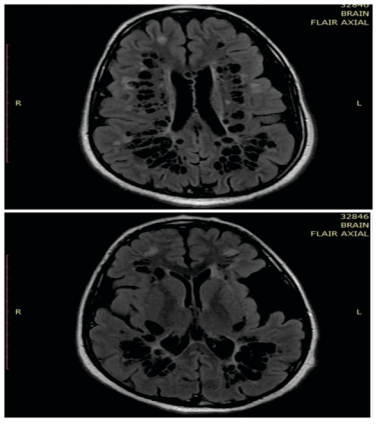
We would take this oppurtunity to present our case series as a pictorial essay . We presentall positive imaging findings we encountered in our cases for easy understanding and accurate image interpretation to aid diagnosis.

PERIVENTRICULAR LEUKOMALACIA (END STAGE) :



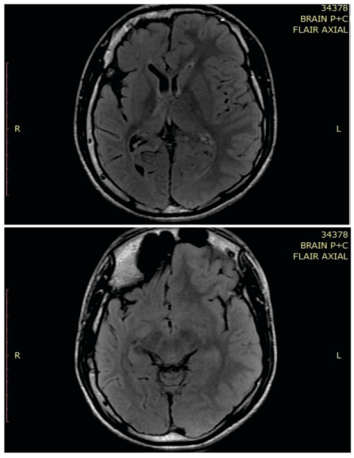
MRI FLAIR images of brain of a child: reveal abnormal severe dilatation of bilateral lateral ventricles, large multiple cystic encephalomalacic changes in the periventricular white matter with involvement of cortex, paucity or thinning of white matter observed bilaterally in keeping with end stage periventricular leucomalacia.

HYPOMELANOSIS OF ITO :



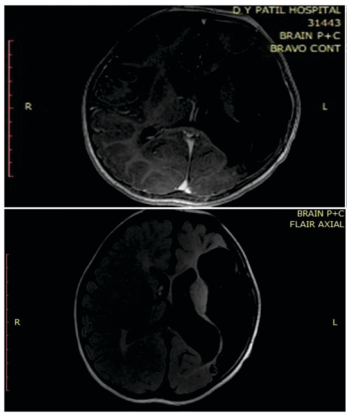
MRI brain FLAIR images of a child: reveal giant perivascular cystic spaces involving bilateral periventricular white matter symmetrically. An arachnoid cyst showing suppression on FLAIR images was also observed in the left anterior temporal region. The patient clinically had hypomelanotic streaks of Blaschko over her upper and lower limbs and back with hypertelorism and impacted dentition diagnostic of a rare phakomatosis called Hypomelanosis of Ito.

DYKE –DAVIDSON-MASSON SYNDROME:



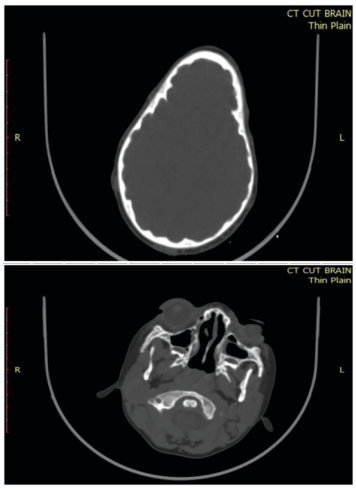
MRI brain FLAIR images of a child: reveal right sided generalised cerebral hemiatrophy with ipsilateral lateral ventricle and sulcal prominence associated with hypertrophy of right sided visualised frontal sinuses and calvarial thickening characteristic of Dyke Davidoff-Masson syndrome.

RASMUSSENS ENCEPHALITIS :



MRI brain FLAIR images in a child: reveal diffuse left sided cerebral hemiatrophy with severe amount of encephalomalacic changes associated with severe left ex vacuo ventricular dilatation, increased FLAIR hyperintensity seen involving the residual cerebral hemisphere on left, no post gadolinium enhancement representing Rasmussen's encephalitis.

CROUZONS SYNDROME:



Non contrast CT images of brain of a child: first image showing increase in size of skull with premature fusion of the sagittal, lambdoid and coronal sutures with resultant increase in the antero-posterior diameter of the skull (Scaphocephaly) with anterior narrowing in the frontal region giving rise to clover leaf shaped skull with multiple areas of scalloping involving the inner table of skull suggestive of copper beaten skull. The CT cuts through the level of maxillary sinuses in the second image reveals bilateral maxillary sinus hypoplasia and bilateral mild degree of exophthalmos representing a hereditary craniosynostosis syndrome called Crouzon syndrome with a triad of skull deformities, facial anomalies, and exophthalmos.

DISCUSSION:

We would be discussing these conditions ie: Periventricular leucomalacia Grade III , Hypomelanosis of Ito , Dyke-Davidon-Masson syndrome , Rasmussens encephalitis , Crouzons syndrome .

Periventricular Leukomalacia:

It is also known as white matter injury of prematurity . It is due to hypoperfusion of white matter which leads to periventricular white matter necrosis followed by cyst formation and finally end stage of encephalomalacic changes, periventricular white matter cysts , paucity of white matter and enlargement of lateral ventricles.80% of patients develop moderate disabilities such as mental retardation , epilepsy and cerebral palsy .MRI findings of periventricular leucomalacia can be divided into early and late stages. Focal or diffuse areas of restriction on Diffusion weighted images which also show corresponding drop on apparent diffusion coefficient values. Late findings of periventricular leucomalacia are those elicited in our case mentioned above ie: periventricular symmetrical cystic changes /encephalomalacic changes, paucity of white matter, predominantly involving the parieto-occipital lobes , adjoining ventriculomegaly of lateral ventricular system and thinning of posterior aspect of the corpus callosum .[1]

Based on the MR findings a scoring system was formulated to evaluate the severity of the hypoxic changes in the brain parenchyma.
Grade 1 - Normal MR findings.

Grade 2 - Areas showing increased signal on long TR sequences.

Grade 3 - Few punctate haemorrhages in the white matter.

Grade 4 - Multiple, more than six areas of punctate haemorrhages in the white matter and/or larger focal haemorrhages but less than 2 in number.

Grade 5 - Extensive changes of increased signal intensities on long TR sequences with areas of punctate or focal haemorrhages.

Grade 6 - Diffuse signal intensity changes with haemorrhagic lesions or cystic lesions involving both periventricular and subcortical white matter [2].

Hypomelanosis of Ito :

This is a form phakomatosis. It is due to decreased melanosome synthesis .It compromises involvement of multiple systems ie: skin hypopigmentation , facial dysmorphism , neurological findings and musculoskeletal abnormalities. Abnormalities involving the skin are characteristic lines of Blaschko which are hypopigmented lines involving the trunk and sparing the soles and palms. [3] Craniofacial abnormailities include macrocephaly, hypertelorism , low set ears and abnormal dentition. MR imaging of brain shows diffuse white matter abnormalities involving periventricular white matter which form abnormal cystic lesions/enlarged perivascular spaces as seen in our case and cerebral atrophy. Rarer features include agenesis of corpus callosum ,lissencephaly , arteriovenous malformations, moyamoya disease.[4] Genetic ,clinical and neuroradiological correlation reach the diagnosis in which neuroimaging plays a pivotal role.

Dyke-Davidoff-Masson syndrome :

This syndrome is composed of a clinical triad of hemiparesis, mental retardation , facial asymmetry and seizures . In literature two types of cerebral hemiatrophy are mentioned by Alpers and Dear, primary and secondary .[5] Hageman et al suggested the cause of primary cerebral atrophy to be lack of cerebral development instead of atrophy . Neuroimaging findings are few typical as shown in the cases above. There is symmetric cerebral atrophy which is most likely due to vascular insult to developing brain during formation or early childhood . Other features are widening of sulci and atrophy of gyri on ipsilateral side, enlargement of ipsilateral lateral ventricle and hypertrophy of ipsilateral frontal sinus. [6]Shen et al .mention three MR imaging patterns of cerebral hemiatrophy: MR imaging pattern I corresponds to diffuse cortical and subcortical atrophy; pattern II corresponds to diffuse cortical atrophy coupled with porencephalic cysts; and pattern III corresponds to previous infarction with gliosis in the middle cerebral artery (MCA) territory..[7]

Rasmussens encephalitis :

Rasmussen encephalitis (RE) is a rare disorder that causes severe chronic, inflammatory disease of the brain parenchyma in paediatric age group usually involving a unilateral cerebral hemisphere .Etiology is unknown. It leads to intractable seizures, cognitive decline and progressive neurological deficits in the affected hemisphere. [8]

There are four stages observed on MRI BRAIN.

Stage I : Presence of abnormal hyperintense T2/FLAIR signal with sulcal effacement indicating underlying cerebral edema.

Stage II : T2 and FLAIR hyperintensies which could be or multifocal involving the cortex or white matter of the hemisphere, predominantly involving insular and peri-insular region.

Stage III: severe unilateral cerebral atrophy with widening of cortical sulci and dilatation of the ipsilateral lateral ventricle.

Stage IV: atrophy of a cerebral hemisphere with significant exvacuo dilatation of adjoining lateral ventricular system suggestive of encephalomalacic changes without abnormal high signal intensity seen involving the abnormal cerebral hemisphere on FLAIR images. Post gadolinium enhancement may or may not be seen.[9]

Crouzons syndrome :

It is also known as craniofacial dysostosis. It is characterised by premature closure of cranial sutures, maxillary hypoplasia, shallow orbits leading to exophthalmos ,midface hypoplasia and bifid uvula. It is distinguished from other craniosynostosis by lack of involvement of hand or foot abnormalities. [10] It is a autosomal dominant disorder due to a mutation in fibroblast growth factor receptor 2 (FGFR2) gene on chromosome 10q25-26 hence genetic screening is essential.[11] Other features include craniocervical junction abnormality , vertebral anomalies commonest being butterfly shaped vertebra , fusion anomalies of vertebra. Complications include hydrocephalus, cosmetic disfiguration, exophthalmos leading to decreased visual acquity. [12] Other clinical features include hypertelorism,beaked nose, short upper lip, mandibular prognathism with no upper and lower limb digital abnormalities. Associations include Chiari I malformation ,stylohyoid ligament calcification , cervical spine fusion anomalies , agenesis of corpus callosum . [13]

CONCLUSION :

Seizures are a common disorder in the paediatric population . We aim to present extremely rare causes of the same .The imaging findings mentioned above are pathognomonic of the disorders hence they aid in their accurate diagnosis working as a sensitive tool for diagnosis and assisting in further management of the patient. Magnetic resonance Imaging is the gold standard of non invasive diagnosis of these rare conditions.

CONFLICTS OF INTEREST :

The authors would like to confirm there are no conflicts of interests present.

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