ORIGINAL RESEARCH PAPER

Neurology

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"STROKE, INTRACRANIAL STENOSIS IN MAYER- ROKITANSKY-KUSTER-HAUSER SYNDROME – FIRST CASE REPORT"

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ABSTRACT

We reported first case of acute ischemic stroke in a patient diagnosed with Mayer Rokitansky Kuster Hauser syndrome. She was a 15 year girl presented with right hemiparesis, headache, and forgetfulness. Her MRI brain revealed acute to subacute large infarct in left middle cerebral artery territory involving left parieto-temporo occipital region and left putamen and left internal capsule. Her cerebral angiography showed total occlusion of left internal carotid artery from its origin. She had primary amenorrhea, developed breast and pubic hairs. MRI pelvis showed absent uterus, absent upper two third of vagina with lower one third of vagina being normally visualized. Both ovaries were normal in position, size, shape and morphology. Solitary malrotated kidney was seen in the pelvic region in midline in the presacral region. Her ANA by immunofluorescence was positive. Patient had no family history. Her karyotyping showed 46, XX suggestive of apparently normal female karyotype. These findings were suggestive of Mayer Rokitansky Kuster Hauser syndrome. Stroke with intracranial vessel stenosis in this syndrome is not reported till now.

BACKGROUND -

Mayer-Rokitansky-Kuster-Hauser (MRKH) syndrome is characterized by aplasia of the vagina with or without concurrent uterine and/or cervical aplasia.

Case Report –

A 15 year girl presented in neurology OPD with sudden onset right hemiparesis, headache, forgetfulness. She also had history of primary amenorrhea. She had no history of chronic medication use or systemic illness in the past. Her developmental milestones were achieved normally and family history was not suggestive of similar illness. She had developed breast and pubic hairs normally. Her MRI brain revealed acute to subacute large infarct in left middle cerebral artery territory involving left parieto-temporo occipital region and left putamen and left internal capsule (Figure 1).

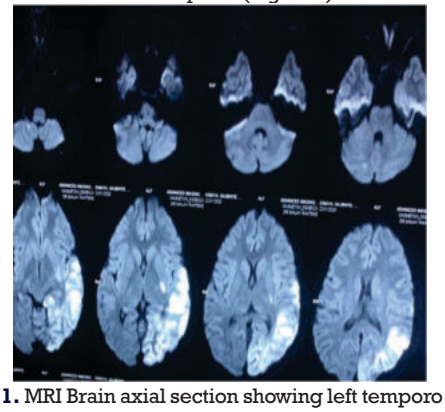

Figure 1. MRI Brain axial section showing left temporo parieto occipital, putamen, internal capsule region large infarct.

Cerebral Angiography showed total occlusion of left internal carotid artery (ICA) from its origin and left anterior cerebral (ACA) and left middle cerebral artery (MCA) and its branches are filling via corticicortical collaterals on right internal carotid artery angiogram (Figure 2).

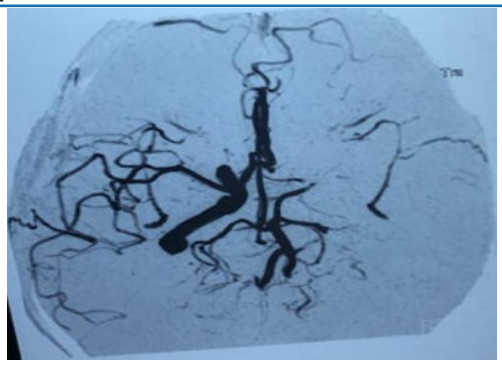

Figure 2. Cerebral Angiography showing total occlusion of left ICA from its origin and left ACA and left MCA and its branches are filling via corticicortical collaterals on right ICA. MRI pelvis showed absent uterus, absent upper one third of vagina with lower two third of vagina being normally visualized. Solitary malrotated kidney was seen in the pelvic region in midline in the presacral region. (Figure 3).

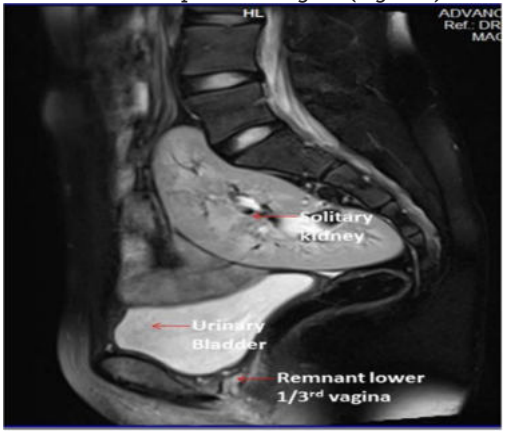

Figure 3. MRI pelvis sagittal section showing absent uterus,

absent upper 1/3rd of vagina with normal lower 2/3rd of vagina and solitary kidney.

Both ovaries were normal in position, size, shape and morphology (Figure 4).

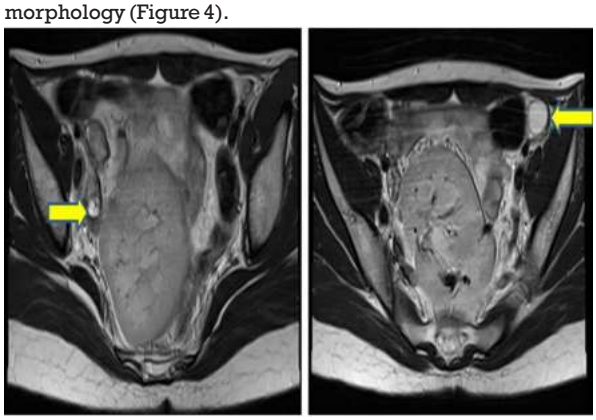

Figure 4. MRI abdomen and pelvis (axial section) showing bilateral normal ovaries.

Her complete blood count was normal and Hb electrophoresis revealed normal AA pattern. Her ANA by immunofluorescence was positive. Her karyotyping showed 46, XX suggestive of apparently normal female karyotype. These findings were suggestive of Mayer Rokitansky Kuster Hauser syndrome. Whole exome sequencing in this patient was normal and no clinically relevant variants were detected.

DISCUSSION -

MRKH syndrome is typically known by the congenital absence of uterus and upper part of vagina in women who have normally developed secondary sexual characters along with a normal female 46, XX karyotype [1]. Ultrasonography is often first diagnostic test in the evaluation of such patients to confirm the presence of ovaries and absence of uterus but results can sometimes be inconclusive [2].However for confirming the diagnosis, MRI is the imaging modality of choice which can also detect associated malformations [3, 4]. The diagnosis of MRKH syndrome imposes a significant psychological burden on patients because of the associated infertility. The distress can be alleviated by psychological counseling and support groups. Treatments include progressive vaginal dilators or surgical creation of a neovagina. Assisted reproductive techniques and surrogacy may be options with regard to fertility [5]. Mayer-Rokitansky-Kuster-Hauser's syndrome with neurological issue and spinal blood vessels malformations has been described [6].Uterine transplant is new procedure may be an option for infertility for patient with MRKH [7].To our knowledge young stoke and intracranial vessel narrowing as presentation of MRKH syndrome is rarely reported in literature.

CONCLUSION -

In young female patient presenting with stroke, intracranial stenosis and primary amenorrhea, probability of MRKH should be kept in mind.

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