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Rare Vasculitis in Childhood-Juvenile Polyarteritis Nodosa

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ABSTRACT: Polyarteritis nodosa (PAN) was first described by Kussmaul and Maier in 1866 .It is a rare fatal vasculitis in childhood, a monogenic auto inflammatory condition caused by mutations in CECR1gene with autosomal recessive inheritance. The disease varies in its presentation from a relatively benign cutaneous form, which may resolve without treatment, to a severe systemic and has an estimated annual incidence of 2.0–9.0/million in adults. It is a systemic necrotizing vasculitis affecting small-medium sized arteries and affecting the multiple organ systems .The onset of the disease is typically between the ages of 25 and 50 years, but despite the lack of good epidemiologic data, it is becoming increasingly apparent that polyarteritis nodosa occurs rather more frequently during childhood. System involvement in polyarteritis nodosa varies, but the skin, the musculoskeletal system, the kidneys, and the gastro-intestinal (GI) tract are prominently affected. Constitutional symptoms like weight loss, abdominal pain, and loose stools are common and suggest mesenteric arterial inflammation and ischemia. Reno vascular arteritis can cause hypertension, haematuria or proteinuria. 50% present with haemorrhagic/ischemic stroke .Cutaneous manifestations include livedo-reticularis, ulceration etc. Since many of the presenting features of poly arteritis nodosa are nonspecific and mimic infections, more common chronic inflammatory diseases of childhood, there is a considerable delay in diagnosis and a consequent delay in starting of treatment, resulting in irreversible end-organ damage or death. The histopathology changes of polyarteritis nodosa are fibrinoid necrosis of the walls of medium or small arteries, with a marked inflammatory response within or surrounding the vessel wall.

11year old male child born by non-consanguineous marriage, by invitro -fertilization who was apparently alright before 5months, started developinganorexia, loose stools with significant weight loss. Patient had melena 3 months back which lasted for one and half months. Family history was significant, mother had two spontaneous abortions at 10 weeks and was on treatment for infertility. Routine investigations were normal, oesophago-gastro-duodenoscopy (OGD), colonoscopy was -suggestive of antral gastritis with multiple non-circumferential ulcers with inflammatory marginsinvolving left colon, treated with probiotics, antibiotics and above mentionedsymptoms resolved.

After two months child was -admitted to our hospital with epistaxis, diplopia, pigmented lesion over legs and hypertension with features of Posterior Reversible Encephalopathy Syndrome, treated as per standard hospital protocol, evaluated for the causes and were normal. Renal function test and renal imaging and Doppler, Echocardiography, MRI angiography, CT aortography was done and was normal. Blood sugar levels and thyroid function test was normal and ANA was negative. Koch's work up was done was normal. Skin biopsy on pigmented lesions of legs was done suggestive of ichthyosis .Child was discharged on oral anti-Hypertensives. After a week the child re-admitted with quadriparesis with bowel bladder involvement, MRI spine was suggestive of hyper intensities involving C4 to D1 with adjacent cord swelling and effacement of thecal sac, most likely vasculitis .Child was suspected to have juvenile Polyarteritis Nodosa, started on steroids and cyclophosphamide. Child received intravenous steroids and cyclophosphamide and shown mild improvement in bilateral upper limb movements and activities. Over the hospital course, child deteriorated and intubated due to respiratory distress and poor respiratory drive, developed ventilator-associated pneumonia (VAP). Inspite of

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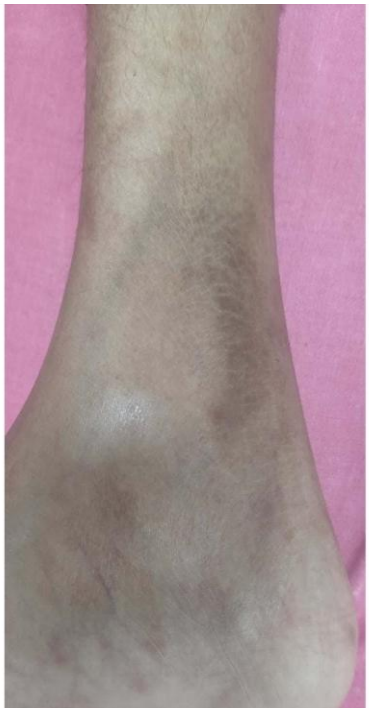
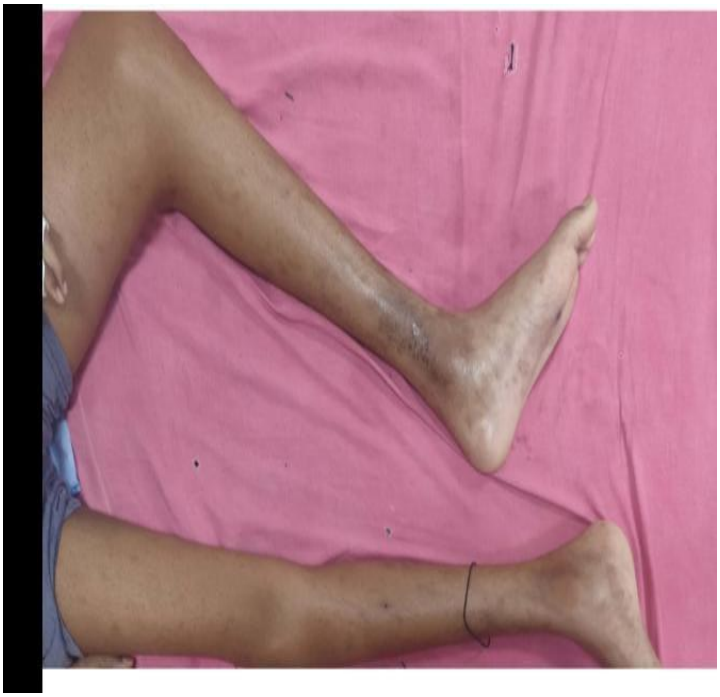
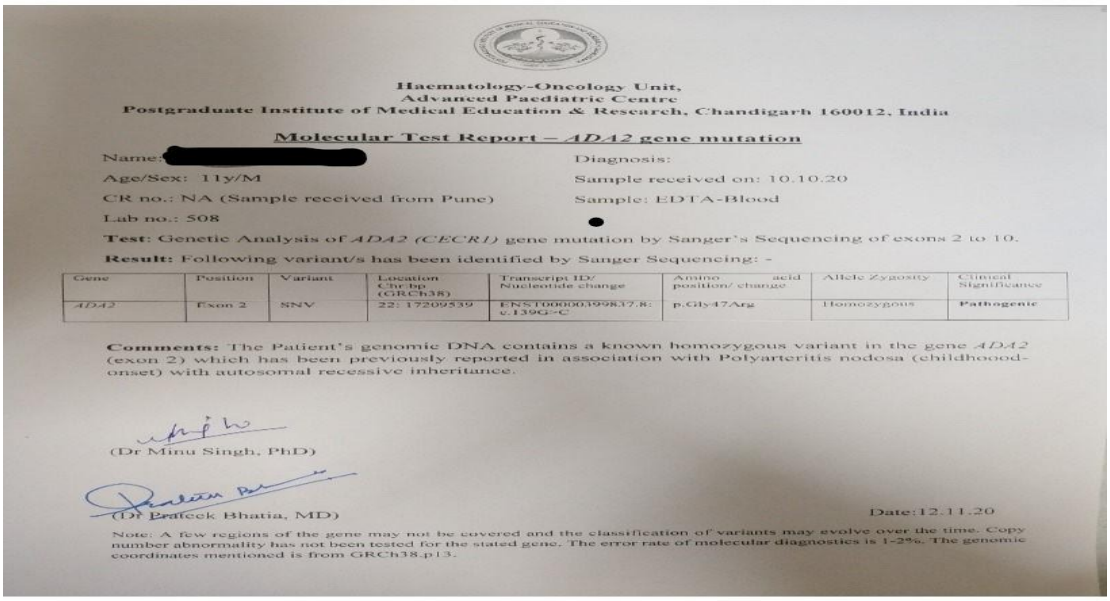
intensive care and treatment, child succumbed due to septicaemia and septic shock before the consideration of Anti TNF-alpha treatment. Later the index patient was positive for known hotspot pathogenic mutation for DADA2.

CONCLUSION: The clinical findings and progression of PAN in childhood is highly variable and may be missed when presented with few nonspecific symptoms at the beginning without definite diagnostic signs, which may appear gradually as the disease progresses. Therefore a high suspicion and thorough clinical examination for any systemic or cutaneous finding may assist in the early diagnosis of such cases.







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Molecular Test Report – *ADA2* gene mutation

Name: [redacted] Diagnosis:
Age/Sex: 11y/M Sample received on: 10.10.20
CR no.: NA (Sample received from Pune) Sample: EDTA-Blood
Lab no.: 508

Test: Genetic Analysis of *ADA2 (CECR1)* gene mutation by Sanger's Sequencing of exons 2 to 10.
Result: Following variant/s has been identified by Sanger Sequencing: -

Gene	Position	Variant	Location Chr: bp (GRCh38)	Transcript ID/ Nucleotide change	Amino acid position/ change	Allele Zygosity	Clinical Significance
ADA2	Exon 2	SNV	22: 17209539	ENST00000399817.8: c.139G>C	p.Gly47Arg	Homozygous	**Pathogenic**

Comments: The Patient's genomic DNA contains a known homozygous variant in the gene *ADA2* (exon 2) which has been previously reported in association with Polyarteritis nodosa (childhoood-onset) with autosomal recessive inheritance.

(Dr Minu Singh, PhD)

(Dr Prafeek Bhatia, MD) Date:12.11.20

Note: A few regions of the gene may not be covered and the classification of variants may evolve over the time. Copy number abnormality has not been tested for the stated gene. The error rate of molecular diagnostics is 1-2%. The genomic coordinates mentioned is from GRCh38.p13.