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SPECTRUM OF AV MALFORMATIONS IN PAEDIATRICS

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Abstract

Cerebral arteriovenous malformations (AVMs) present a challenge to diagnose in children with developmental disability, because of the overlap in behavioral symptoms and neurologic manifestations. A 16 year old male child came with complain of progressive dyspnoea since early childhood with recurrent epistaxis and had severe anaemia on admission. On detailed investigations and work up pulmonary venous malformations were found and genetic study for HHT was planned. Few more cases with the similar spectrum of disorder have been covered.

Keywords: Cerebral arteriovenous malformations, Paediatric, developmental disability.

INTRODUCTION

Paediatric arteriovenous malformations (AVMs) are uncommon vascular anomalies that are characterised by fistulous connections between arteries and veins without a healthy intervening capillary bed. They present a challenge to diagnose in children with developmental disability, because of the overlap in behavioral symptoms and neurologic manifestations. A 16 year old male child came with complain of progressive dyspnoea since early childhood Shunting of arterial blood and increased pressure on the venous vessels may cause the AVM to become larger and perhaps rupture, resulting in either tiny or massive intracranial bleeding. The extent of the AVM's subsequent bleeding and its location influence the neurologic consequences that follow. Large AVMs can occasionally obstruct the flow of cerebrospinal fluid, which can lead to hydrocephalus and elevated intracranial pressure.[1]

The prevalence rate in children as well as adults is believed to be between 1 to 10.3 per 100,000,[2] and the majority of cases are asymptomatic or discovered by accident during neuroimaging.[3] Children with AVMs are more likely than adults to present with bleeding, and fatality rates from bleeding have been recorded to reach 25%.[4] Medical emergency can be avoided, and interventions are more likely to be successful, if an AVM is discovered before a significant haemorrhage. In this report we present case reports of pediatric patients who presented with an AVM.

CASE REPORT

Clinical Description

Case 1

A 16 year old male; 2nd born of NCM r/o Indore came with complain of progressively increasing dyspnea since 1 week. Significant night time awakening due to breathlessness, easy fatiguability, not thriving well and mild circumoral cyanosis occasionally noticed since early childhood.

No h/o oedema, arthralgia, arthritis, rashes and no other bleeding manifestations, no history s/o CHD, no h/o chronic diarrhoea or prolonged fever.

Vitals- GCS: E4V5M6, Sick Looking, Irritable. HR: 150 -160/min, RR: 36 - 40/min, PP: Bounding, Regular, No Radio-Radial or Radio-Femoral Delay, Temp: 98.8 C, Pallor marked, Clubbing: Grade 3, Cyanosis + over lips, Mild pedal edema +, P/A: Liver palpable 4 cm BCM; span 11 cm, soft, mild tenderness in right hypochondrium;

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margins normal, R/S: Air entry equal; use of accessory muscles seen, CVS: Hemic murmur +; JVP raised, SMR: Pre pubertal(axillary hair-absent; pubic hair-sparse), Weight: 40 kg (3 to 10th P), Height: 145 cm (3 to 10th P), BMI: 19.2 kg/m sq (10 to 25 P), Inference: Patient had delayed growth spurt and had stunted growth.

Differentials:

- Idiopathic Pulmonary Hypertension with chronic anemia? Nutritional (dimorphic)? Anaemia of chronic illness
- ? Pulmonary venous malformation with? Hereditary hemorrhagic Telangiectasia

Diagnosis

- **Investigations**: CBC: 2.8 HB; WBC and plt –wnl, PS- normocytic normochromic, Sr.Protein: 5.15/2.73/2.42/1.1, Stool r/m: WNL, Urine r/m: WNL, Chest x ray: homogenous opacity in right lung, Retic count: 0.4 % (0.15), RFT: WNL, LFT: WNL, HIV: NR, HRCT CHEST- Dense areas of consolidation in the anterior segment of both upper lobes s/o Infective Etiology, Dilated serpentine enhancing lesions adjacent to consolidations ?Pulmonary Varices, Contrast ECHO: Pulmonary AV malformation, Conventional Pulmonary

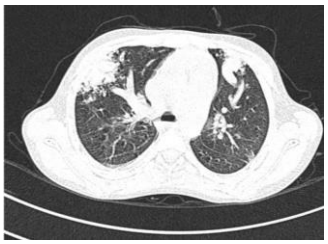

Figure 1. CT Angiography: Multiple significant Pulmonary Arterio-Venous Malformations

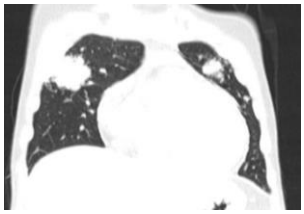

Figure 2. Conventional Pulmonary Angiography: Multiple significant Pulmonary Arterio-Venous Malformations

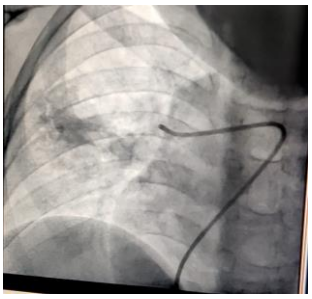

Figure 3. Contrast ECHO: Pulmonary AV malformation

Patient outcome:

Patient tolerated the procedure well, Vitals were stable. Post procedure he was started on aspirin and metoprolol. On follow up there was significant improvement in dyspnea and oxygen saturation 97% on room air, weight gain is present and genetic work up for osler weber rendu is being planned.

PAVMs are the abnormal communications b/w pulmonary arteries & veins. Blood bypasses normal oxygen-exchanging pulmonary capillary bed returning desaturated blood to the pulmonary veins. Return of desaturated blood to the pulmonary veins becomes significant measurable arterial oxygen desaturation and cyanosis results. The age at the first presentation ranges from newborn to 70 years but the majority of cases are diagnosed in the first three decades of life.

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A. Before treatment **B. After treatment**

Figure 4. Oxygen saturation

Hereditary hemorrhagic telangiectasia: Presence of multiple arteriovenous malformations (AVMs) that lack intervening capillaries and result in direct connections between arteries & veins.The most common clinical manifestation is spontaneous and recurrent epistaxis beginning on average age of 12 years. Pulmonary AVMs occur in approximately 30%-50% of affected individuals and can develop over time but rarely after age 30. Epistaxis, mucocutaneous telangiectasia, and GI bleeding are most common presentations. A multidisciplinary approach is needed to evaluate the many potential manifestations of HHT to achieve a complete diagnostic picture. Patients with P-AVM in particular, as in this patient, are at serious risk of complications due to paradoxical embolism, indicating the need for early diagnosis and intervention

Clinical diagnosis of HHT is based on the Curaçao diagnostic criteria:

- History of recurrent epistaxis
- Multiple telangiectasias (Lips, Oral cavity, Nose)
- Involvement of visceral lesions (Lung, Liver, Brain)
- First-degree relative with HHT

Diagnosis is considered "definite" if 3 or more criteria are present, and "suspected" if 2 are present.

80% of people who meet the clinical diagnostic criteria for HHT are found to have a mutation in either the ENG (HHT type 1) or ACVRL1 (HHT type 2) gene. 3-5% of clinically diagnosed individuals test positive for a mutation in the SMAD4 gene, which causes a combined syndrome of HHT and Juvenile Polyposis. About 10-15% of patients will not have a mutation detected in a known HHT gene, and in these cases a diagnosis is made based on clinical evaluation alone. PAVMs are present in up to 60% of children with HHT. Most PAVMs can be detected on initial screening with TTCE, but nearly 30% of children with HHT can develop new PAVMs over time. Furthermore, up to 25% of children initially diagnosed with small PAVMs can develop larger PAVMs requiring embolization. Nearly 20% of those children who had successful embolization therapy needed repeat intervention. Children diagnosed with HHT need continued assessment for PAVMs throughout childhood. Brain abscess, Hemorrhagic or ischemic stroke, High-output congestive heart failure, Chronic GI bleeding, Portal hypertension with esophageal varices, Pulmonary hemorrhage, Liver cirrhosis

Of patients who have pulmonary AVMs, 2% per year are estimated to have a stroke, and 1% per year are estimated to develop a brain abscess

Case 2

A 7yr old female child presented with complain of easy fatiguability and failure to gain weight with no past history of hospital admission or IV antibiotics or history of squatting. Child was maintaining spo2 0f 82% on O2 support with cyanosis and pandigital clubbing with normal systemic examination.

Investigations:

- 2D Echo – normal
- Contrast ECHO _ s/o pulmonary AV malformation
- CT pulmonary angio and abdomen – Extra hepatic Portal vein to IVC shunt (Abernathy malformation type 1b shunt)

Multiple hepatic lesions like FNH/regenerative nodules due to altered hepatic vasculature

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Discussion:

Hepato- pulmonary syndrome

May be asymptomatic, well up to adulthood

Features related to shunting of portal blood like hepatopulmonary syndrome, hepatic encephalopathy, metabolic dysfunction, associated congenital anomaly and features secondary to hepatic lesion

Treatment:

- Type 1- Liver transplant
- Type II- Closure of shunt (intervention/surgical)

Case 3

8 yr old male child presented with complain of easy fatiguability and effort intolerance, was hemodynamically stable and had cyanosis and generalised pan digital clubbing on examination

Investigations:

- 2D Echo – normal
- Contrast Echo – s/o Pulmonary AV malformation
- Adv for CT Pulmonary angiography

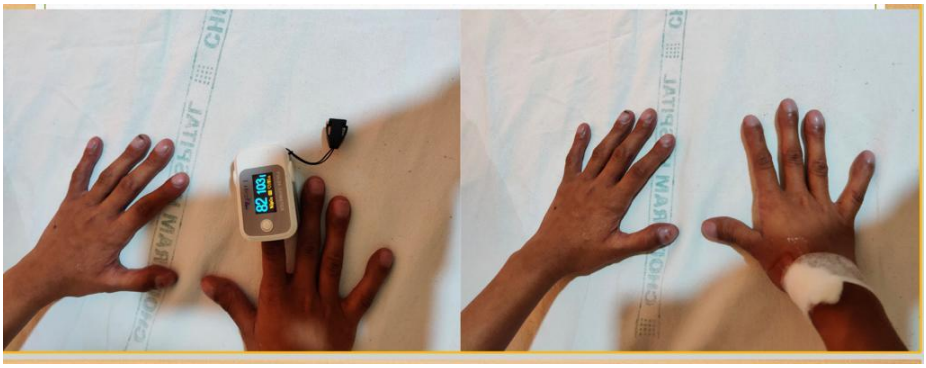

Figure 5. Clubbing and cyanosis

Case 4:

A 5 yr old male child presented with shortness of breath and effort intolerance. Child had saturation of 88% with tachycardia and no significant finding on general and systemic examination

Investigations:

- 2D ECHO – normal
- Contrast ECHO – Pulmonary AV malformations

Adv-

- CT pulmoangiography
- Contrast ECHO _ s/o pulmonary AV malformation

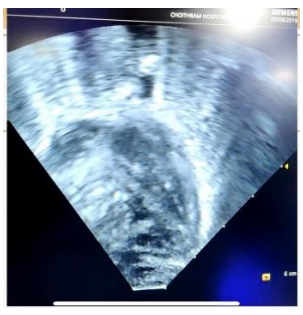

Figure 6. Contrast 2 D ECHO showing AV malformation

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- CT pulmonary angio and abdomen – Extra hepatic Portal vein to IVC shunt (Abernathy malformation type 1b shunt)
- Multiple hepatic lesions like FNH/regenerative nodules due to altered hepatic vasculature

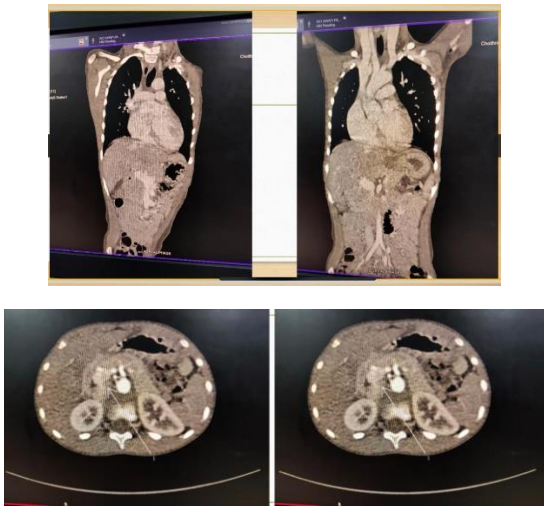

Figure 7. CT pulmonary and abdomen angiography

Discussion:

Hepato- pulmonary syndrome

May be asymptomatic, well up to adulthood

Features related to shunting of portal blood like hepatopulmonary syndrome, hepatic encephalopathy, metabolic dysfunction, associated congenital anamoly and features secondary to hepatic lesion

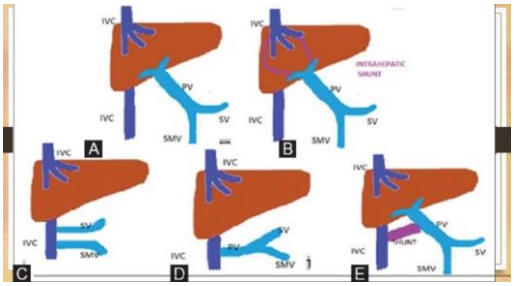

Figure 8. Hepato portal shunts

Treatment:

- Type 1- Liver transplant
- Type II- Closure of shunt (intervention/surgical)

Case 5:

8 yr old male child presented with complain of easy fatiguability and effort intolerance, was hemodynamically stable and had cyanosis and generalised pan digital clubbing on examination

Investigations:

- 2D Echo – normal
- Contrast Echo – s/o Pulmonary AV malformation

Adv for CT Pulmonary angiography

Case 6

A 5 yr old male child presented with shortness of breath and effort intolerance. Child had saturation of 88% with tachycardia and no significant finding on general and systemic examination

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Investigations

- 2D ECHO – normal
- Contrast ECHO – Pulmonary AV malformations

Adv- CT pulmoangiography

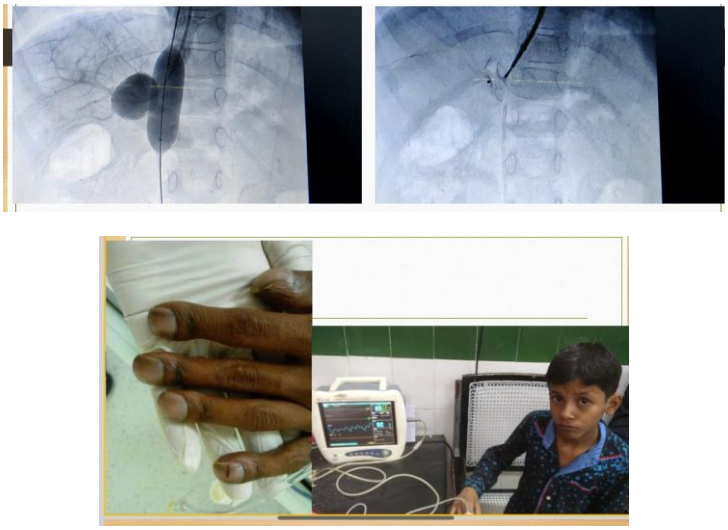

Figure 9. AV malformation

Case 7

A 1 day full term/ AGA newborn with tachypnoea and desaturation referred for further evaluation and management. Admitted to NICU with initial stabilisation done acc. to protocol. Baby had tachycardia and was maintaining 75% Spo2 on o2 support

I*nvestigations*

- 2D ECHO – s/o pulmonary atresia, PDA supplying pulmonary artery, IV prostin started after which saturation improved and was planned for PDA stent thereafter

Case 8

A 3 yr old 9kgs female child with c/o recurrent respiratory infection and multiple episodes of hospitalisation referred for cardiac evaluation. On examination child was maintaining saturation on rom air, had tachycardia and tachypnoea with SCR+, hyperdynamic precordium and apex. On auscultation child had S1 – normal, S2 wide split, ESM grade IV/VI at left upper parasternal border, PSM +

Investigations

- 2D ECHO – s/o 6mm peri membranous VSD with moderate to severe MR
- Significant LA/LV volume overload and PAH

Advised for VSD D? C

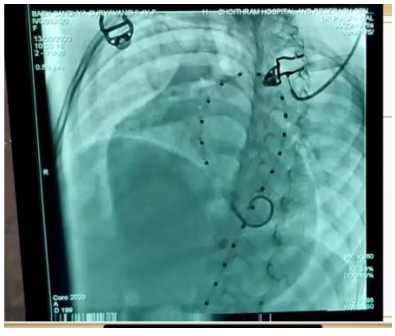

Figure 10. VSD device closure

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Figure 11. After treatment

RESULT:

Of all the clinical cases in our study it was found that incidence of symptoms in children with AV malformation were in the following order

- Easy fatiguability (66 %)
- Recurrent infections (50%)
- Not thriving well (50 %)
- Recurrent epistaxis (33%)

CONCLUSION

AV malformation is a rare disorder. Though children presenting with clinical features of progressive dyspnoea, not thriving well, recurrent infections and epistaxis with clubbing and cyanosis on general examination in which 2 D ECHO finding are non-conclusive, a possibility of AV malformation should always be ruled out so and further genetic evaluation should be done for better outcome of the child

ACKNOWLEDGEMENT

None

DECLARATIONS

Conflicts of interest

There is no conflict of interest with publication of manuscript or an Institution or product that is mentioned in the manuscript and/or is important to outcome of study presented.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent.

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REFERENCES:

1. Baskaya MK, Jea A, Heros RC, Javahary R, Sultan A. *Clinical Neurosurgery.* Vol. 53. Lippincott Williams & Wilkins; 2006. Cerebral Arteriovenous Malformations; pp. 114–115.
2. Whigham KB, O'Toole K. Understanding the Neuropsychologic Outcome of Pediatric AVM within a Neurodevelopmental Framework. *Cog Behav Neurol.* 2007;20:4.
3. Millar C, Bissonnette B, Humphreys RP. Cerebral Arteriovenous Malformations in Children. *Can J Anaesth.* 41(4):321–331.
4. Abdalla SA, Letarte M. Hereditary haemorrhagic telangiectasia: current views on genetics and mechanisms of disease. J Med Genet. 2006;43:97–110.
5. Bayrak-Toydemir P, McDonald J, Akarsu N, Toydemir RM, Calderon F, Tuncali T, Tang W, Miller F, Mao R. A fourth locus for hereditary hemorrhagic telangiectasia maps to chromosome 7. Am J Med Genet A. 2006a;140:2155–62
6. Bayrak-Toydemir P, McDonald J, Markewitz B, Lewin S, Miller F, Chou LS, Gedge F, Tang W, Coon H, Mao R. Genotype-phenotype correlation in hereditary hemorrhagic telangiectasia: mutations and manifestations. Am J Med Genet A. 2006b;140:463–70.