IOSR Journal of Dental and Medical Sciences (IOSR-JDMS)
e-ISSN: 2279-0853, p-ISSN: 2279-0861.Volume 17, Issue 2 Ver. 10 February. (2018), PP 46-48
www.iosrjournals.org

OSLER- WEBER -RENDU Disease Presenting As Recurrent Pulmonary Arteriovenous Malformations

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Abstract: *Osler-Weber-Rendu Syndrome Is A Rare Genetic Vascular Disorder With Various Clinical Manifestations. Most Common Manifestations Were Epistaxis And Gastrointestinal Bleeding With ArterioVenous (AV) Malformations Also Occurring In Pulmonary And Hepatic Circulation. Management Include Treatment Of Vascular Lesions. Our Case Report Describes A Case Of Hereditary Hemorrhagic Telangiectasia (HHT) Of An Adult Male, Associated With Recurrent A-V Malformations (AVM's) In Lungs Who Presented With Recurrent epistaxis And hemothorax And Resulting In Breathing Difficulty.He Was Diagnosed With A-V Malformations In Lungs Twice Within 9 Years. On Detailed Evaluation He Was Diagnosedwith Osler‐ Weber‐ Rendu Disease Based OnCuracaocriteria. We Successfully Managed the Patient's Hemothorax by Thoracentesis. This Case Emphasizes The Need For Careful Examination And Investigation And To Consider Such Rare Diseases When All The Common Causes Of Hemothorax Are Ruled Out.*

Key Words*- AV Malformation, Epistaxis,Hereditary Hemorrhagic Telangiectasia*

Date of Submission: 09-02-2018 Date of acceptance: 23-02-2018

I. Introduction

Osler-Weber-Rendu Syndrome Also Known As Hereditary Hemorrhagic Telangiectasia (HHT) Is A Rare Genetic Autosomal Dominant Disorder.The Typical Findings Of The Disease Are Telangiectasia In Skin And Mucous Membranes, And AVM'sPresenting In The Organs Like Lung, Intestine, Brain And Liver, Recurrent Epistaxisand With Familial Occurrence[1].The Prevalence Of This Disease Is 1 In 5000-8000 Population. However, The Prevalence May Be Underestimated Because Many Cases May Be Asymptomatic[2].OnsetOf Symptoms May Be Delayed Until The Fourth Decade Of Life (~90% Of Patients Manifest By Age 40 Years) Or Later[3,4,5].The Vascular Malformations In HHT Consist Of Direct Arteriovenous Connections Through Thin Walled Aneurysms, And Range From Small Telangiectases (Predominantly Mucosal, Skin, And Visceral) To Larger Visceral AVM[6].Most Clinical Complications Of The Disease Are Related To Pulmonary AVM (PAVM) About 30-60% [7,8]. Treatment Is Supportive And Helps Prevent Complications.

II. Case Report

This Is A Case Of 52year Old Obese Male,Known Smoker Came To Our Hospital With Complaints Of Breathlessness (NYHA Grade 3),Which Increased On Lying Down Position(Orthopnoea) ,Paroxysmal Nocturnal Dyspnea For Last 20 Days And Left Sided Chest Pain Which Was Moderate In Nature And Bilateral Pedal Edema Since Last 3 Months.HeHad An Episode Of Epistaxis One Month Backwhich Settled With Conservative Treatment.

HeWas Previously Admitted In Local Hospital For The Above Complaints, Investigations showed Hemoglobin-13.7g/Dl, Total Leukocyte Count-18,400 Cumm,Platelet Count-1 Lakhs/Cumm, Creatinine-1.78mg/Dl,SerumBiluribin-1.3mg/Dl.ECG Showed Inverted T Waves In Precordial Leads.He Was Brought To Our Hospital On Vasopressor Support For Further Management.

He Is A Known Case Of Right Pulmonary Artery A-V Fistulaand Had Right Middle And Lower Lobectomy 6 Years Back.There Was No History Of Pulmonary Tuberculosis, Diabetes Mellitus And Hypertension. On Enquiring It Was Found That His Mother,Sister And Brother Had A History Of Telangiectasia But There Was No History Of Epistaxis In Any Of His Family Member.

On Examination, General Condition Sick,He Was Tachypnoeic (RR-28/Min), Tachycardia (PR-110/Min),Saturation-94% (@2lit/Min O_2),Hypotensive(On Vasopressors). Patient Had Clubbing In His Fingers,Telangiectasia All Over Body But Mainly On His Chest And His Back.He Also Had Pedal Edema In Both Legs.There Was No Cyanosis,Lymphadenopathy Or Icterus. Auscultation Revealed Decreased Vesicular

Breath Sound In Both Lower Lobes With Bilateral Crepts.Rest Of The Results Of The System Examination Were Normal.Treatment Was Started In The Form Of Empirical Antibiotics, Nebulization, Non-Invasive Ventilationand Other Supportive Measures.Labs: NTPROBNP-76.49; TROP I<0.16;Lactate-1.1.LFT Was Within Normal Limit.Creatitine-1.1mg/Dl.His Coagulation Profile Was Normal. Chest X-Ray Showed Left Lower Zone Effusion With Blunting Of Costophrenic Angle With Right Lower Lobectomy Status

2D ECHO Showed Normal LV Size,No Regional Wall Abnormality,LVEF-60%,Normal Right Ventricularsize,No Pulmonary Artery Hypertension,CVP-5,CO-4.6lit/Min,SVR-1095d With Left Pleural Effusion.Gradually Patient Improved,Breathlessness Decreased, His Vasopressor Requirement Was Decreased, Type 2 Respiratory Failure Also Improved,So He Was Shifted Out Of ICU To HDU.Blood, Sputum And Urine Culture Were Sterile. Patient Improved Symptomatically So He Was Shifted To Ward.

Later,Patient Had Symptoms Of Breathlessness,Chest Pain And Had Desaturation,So He Was Again Shifted To HDU.TROP I -Negative,ECG Showed Sinus Tachycardia,Chest X-Ray Done Showed Increased Pleural Effusion And Blunting Of Left Costophrenic Angle.USG Guided Pleural Fluid Aspiration Was Done And 850ml Of Hemorrhagic Fluid Was Aspirated.Pleural Fluid WBC Count -2240,RBC Count -1320000 Suggestive Of Hemothorax.Differential Leucocyte Count Showed –Neutrophils 40%,Lymphocytes 60%.Pleural Fluid Glucose -102mg/Dl,Protein-5.3,LDH-6920.Gene Xpert Tb- Negative.Aerobic Culture Showed No Growth.Pleural Fluid Hematocrit -13.1

CT Pulmonary Angiography Was Done To Further Look For The Source Of Bleeding Which Showed A Hyper Vascular Nodular Lesion Of 11mm Size Seen In Left Upper Lobe,Placed Peripherally With A Feeding Pulmonary Artery Branch And A Draining Pulmonary Vein,Suggestive Of Pulmonary A V Fistula.The Linear Filling Defect Is Also Seen In The Distal Left Upper Lobe Pulmonary Artery Branch In The Anterior Segment Suggestive Of Thrombus.Subtle Patchy Wedge Shaped Opacities Are Seen In The Peripheral Left Upper Lobe Suggestive Of Pulmonary Infarcts.

Rheumatology Scree-(RF,ANA,P-ANCA,C-ANCA) Was Negative.Patient Was Taken For Angiography And Later Thromboembolisation Was Doneand Was Discharged In Stable Condition And Is On Regular Follow Up. Our Case Was Unique As We Got All The Four Criteria –Epistaxis, Telangiectasia,A-V Malformation And Family History For The Diagnosis Of Osler Weber Rendu Disease.

III. Discussion

Osler–Weber–Rendu Syndrome Also Known As Hereditary Hemorrhagic Telangiectasia (HHT) Is An Autosomal Dominant Vascular Disorder Which Affects Multiple Systems[11]. The Classic Clinical Triad Includes Recurrent Epistaxis, MucocutaneousTelangiectasias, And One Affected First-Degree Relative[12].MutationsIn Genes, Endoglin On Chromosome 9(HHT Type 1) And Activin Receptor-Like Kinase-1 On Chromosome 12 (HHT Type 2), Are Associated With HHT. Curacao Diagnostic Criteria Is Used For Diagnosis Of HHT. It Is Based On Four Findings. Epistaxis: Spontaneous, Recurrent Nose Bleeds, Telangiectasias: At Characteristic Sites (E.G. Lips, Oral Cavity, Fingers And Nose), Visceral Lesions: Gastrointestinal Telangiectasias (With Or Without Bleeding) And Arteriovenous Malformations (Pulmonary, Hepatic, Cerebral And Spinal), Family History: A First-Degree Relative With Hereditary Hemorrhagic Telangiectasia. Diagnosis Is Definite If 3+ Criteria Are Present, Suspected If 2 Are Present And Unlikely If Only 1 Criteria Is Present[11].

HHT Generally Develops With Increasing Age With Epistaxis Being The Earliest Sign Which Often Occur In Childhood. Pulmonary AV Malformation Generally Develop After Puberty. Patients Can Have No Symptoms, Or They Can Present With Bleeding, Such As Hemoptysis Or Massive Thoracic Haemorrhage[11]. Up To 70% Of Patients With HHT Tends To Develop Hepatic Avms, Which Are Often Silent. Cerebral Avms Are Present In More Than 10% HHT Individuals. TelangiectasesOf The Skin And Buccal Mucosa Are Quite Common And Occur In About 75% Of Individuals[2].

Multidisciplinary Collaborative Approach Is Required For Management[1]. HHT Should Be Suspected In All Patients With Pulmonary AVM's, Early Onset Stroke Or Brain Abscess[2]. Epistaxis Should Be Managed By Local Therapies Like Saline Sprays And Nasal Humidification For Prevention. For Recurrent Bleeds Laser Therapy And Other Surgical Interventions Can Be Used. Pulmonary AVM's Are Treated By Embolization (Treatment Of Choice) And Indicated When Feeding Artery Diameter Is Atleast 3 Mm. Surgery Is Done In Patients Who Can't Undergo Embolization (E.G. If The Pulmonary Malformations Are Not Amenable To Embolization And Untreatable Contrast Allergy)[11]. Family Screening Should Be Done For Early Detection Of HHT. Prognosis Of The Disease Is Quite Variable And Depends On The Disease Manifestations And Their Management.

IV. Conclusion

Osler-Weber-Rendu Syndrome Is A Rare Genetic Disease With Variable Presentation. A High Index Of Suspicion Is Needed For Diagnosis. Management Includes Early Detection Of Organ Involvement And Treating Manifestations And Measures To Prevent Further Recurrence.

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