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Comprehensive Case Analysis of Primary Pulmonary Hypertension in a Middle-Aged Male: Clinical Evaluation, Genetic Screening, and Management Strategy

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Abstract: ***This case offers a striking example of how primary pulmonary hypertension (PPH) can present subtly yet demand a multi-layered diagnostic and therapeutic approach. A 40-year-old male, euglycemic, normotensive, and without a history of smoking or asthma, reported a month-long history of dyspnea on exertion without associated palpitations, syncope, or orthopnea. It is evident that the absence of common cardiovascular red flags initially could mask the seriousness of the underlying pathology. Clinical evaluation and imaging revealed right atrial and ventricular dilation, moderate pulmonary arterial hypertension, and preserved left ventricular function, with no evidence of shunting or valvular disease. Genetic screening for BMPR2, CAV1, KCNK3, EIF2AK4, ALK1, ENG, and SMAD9 genes was conducted to assess hereditary predisposition, underscoring the forward-looking approach to management. This suggests that early genetic evaluation, alongside conventional imaging and hemodynamic studies, is pivotal in refining prognosis and guiding treatment. Therapeutic intervention included sildenafil citrate, bosentan, digoxin, and frusemide, with advisement for possible heart or lung transplantation. The case stands as a reminder that timely recognition and a layered strategy—blending pharmacotherapy, genetic insight, and surgical foresight-can meaningfully influence outcomes in complex cardiopulmonary disorders.***

Keywords: primary pulmonary hypertension, genetic evaluation, right heart dysfunction, clinical case study, targeted therapy

Case Study

40 Years old male Euglycemic, normotensive, Non Asthmatic, Non Smoker, Alcoholic

Presented with C/o DOE – 1 Month
No C/o Palpitation / Orthopnoea / PND
No H/o of Syncope / Pre syncope / Giddiness
No H/o hematemesis / hemoptysis
No h/o CVA / CKD / MI / BA / TIA

O / E – Heart Rate 80/mt
SPo2 99%
BP 110 / 70 mmhg
CVS – S1S2+
EDM +
P2 Loud
RS – Clear

Hb - 16 gm TC - 179

FBS - 101 HDL - 41
PPBS - 142 Non HDL - 138
Urea - 34 LDL - 123
Creatinine - 0.9 TGL - 72
Na - 142 TB - 0.8
K - 3.8 SGOT - 28
TSH - 3.7 SGPT - 26
Uric Acid - 7.6

EC - NSR
RAD
CCW Loop
Echo - No RWMA
EF - 58%
Dilated RA & RV
D Shaped LV
RV Dysfunction
IVC – 2.48 cm
Grade I LVDD

CT Pulmonary Angiography

Main Pulmonary artery - 40 mm
Right Pulmonary artery - 26 mm
Left Pulmonary artery - 29 mm

TEE- Dilated RA, RV, Pas
Mild PR
Mild TR
Moderate PAH
No Left to Right Shunt
No ASD

Diagnosis
Primary Pulmonary Hypertension
Gene Test- BMPR2
CAV1
KCNK3
EIF 2AK4
ALK1
ENG
SMAD9

has been evaluated

Treated with
Sildenafil Citrate

Bosentan
Digoxin
Frusemide

and advised Evaluation for possible Heart / Lung Transplant

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