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ACHONDROPLASTIC DWARF WITH AORTIC STENOSIS AND PULMONARY STENOSIS WITH POST CARDIAC INTERVENTION – A SUCCESSFUL OBSTETRIC OUTCOME

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ABSTRACT Achondroplasia, inherited as an autosomal dominant trait, is a genetic disorder of bone growth occurring in 1:10,000 to 1:40,000 in all races and sexes. Management of pregnancy in achondroplastic dwarfs is inherently high risk, due to its adverse effect on obstetrical outcomes, namely, pre-eclampsia, polyhydramnios, respiratory compromise, contracted pelvis etc. Incidence of aortic valve disease and pulmonary valve disease in achondroplastic patients is again very rare, with hardly a handful of documented cases available. We present this case of a 21 year old primigravida with aortic stenosis and pulmonary stenosis, with cardiac symptoms aggravated due to the pregnancy, who underwent a valvuloplasty during her gestational period, culminating in a successful Caesarean delivery.

KEYWORDS : achondroplasia, Caesarean, aortic stenosis, pulmonary stenosis, valvuloplasty

Introduction

Achondroplasia is one of the best known and most common types of nonlethal skeletal chondrodysplasia. The incidence is about one in 10,000 to 30,000 live births[1-3]. It is a genetic disorder and inherited as an **autosomal dominant trait.** These individuals have normal mental and sexual development, and life span may be normal. Information regarding obstetric behavior in achondroplastic females is scarce in literature. However, problems such as pre-eclampsia, polyhydramnios, respiratory compromise, contracted pelvis necessitating lower section caesarean section, prematurity and foetal wastage, etc, have been reported.The condition can be associated with several other congenital anomalies, the commonest being respiratory anomalies; however, few isolated reports of it being associated with heart disease, especially valvular heart defects are also available. Isolated congenital valvular stenosis of either aortic or pulmonary valve is commonly seen yet the presence of both these lesions in the same patient is rare. Isolated pulmonary valve stenosis (PS) accounts for 8–10% and aortic valve stenosis (AS) accounts for 3–8% of patients with congenital heart disease [4-5]. A review of the literature revealed only 15 cases of combined valvular stenosis of [7] aortic and pulmonary valves reported to date [6-11]. The increased importance of combined AS and PS, in addition to its rarity, stems from the fact that failure to recognize and adequately correct both lesions may be catastrophic. We present here a case of 21 year old achondroplastic dwarf with **combined congenital severe valvular aortic stenosis & pulmonary stenosis diagnosed at 18 wks gestation.** The patient underwent successful balloon dilatation of both these valves in the same sitting at 19 wks.Pregnancy was continued till 37 wks and LSCS was done with good maternal & fetal outcome. Because of the rarity of this condition and its adverse impact on obstetrical outcome, this case of achondroplasia with aortic and pulmonary stenosis in pregnancy is worth discussing.

Case report

A twenty one year old married since 3 yrs (husband dwarf) suspected achondroplastic dwarf (height 101 cm, wt 30 kgs) primigravida with four months of amenorrhoea had been referred from other medical college to our obstetrics and gynaecology department for medical termination of pregnancy, in view of coexisting aortic and pulmonary stenosis. From her very childhood she would have episodes of breathlessness, never previously evaluated; those were exacerbated for two months prior to reporting in March this year (last menstrual period being six and a half months back in September 2016). Preliminary tests confirmed pregnancy via urine pregnancy test and serial ultrasonograms were taken confirming a live foetus. On exacerbation of breathlessness, she underwent cardiac evaluation in January 2017 and echocardiography revealed a tricuspid aortic valve with coexisting severe aortic and pulmonary stenoses. She was referred to our hospital immediately with 5 months of amenorrhoea with Systolic thrill palpable at both second right and left intercostal spaces. On auscultation, ejection click with grade 4 systolic ejection murmur was heard over pulmonary area and was diagnosed as being achondroplastic with acyanotic heart disease with severe aortic stenosis and pulmonary stenosis. A multidisciplinary meeting involving obstetrician & cardiologist was held to determine the appropriate management strategy.Pt explained high risk of MTP v/s continuation of pregnancy. Patient insisted for continuation of pregnancy and underwent balloon aortic valvuloplasty and balloon pulmonary valvuloplasty and was discharged with tab atenolol in view of raised heart rate and blood pressure. She first reported to the Gynaecology out patient department in March 2017 wherein she was diagnosed as being achondroplastic with acyanotic heart disease with aortic and pulmonary valuplasty and having intra uterine growth retardation. Upon admission, she was started on IUGR regimen. Serial ultrasonograms were performed between March to May 2017 that showed the presence of a single live intra uterine gestation (34 weeks as on 15th of May). A repeat 2d echo done showed mild aortic stenosis and mild pulmonary stenosis and moderate pulmonary hypertension.Close maternal and foetal surveillance was done. Finally she was taken up for emergency lower segment Caesarean section at 37 weeks; male child of 2.25 kg was delivered at 2.05pm uneventfully and she was kept under observation in the critical care unit for three days. She was discharged after fourteen days.

Discussion

Achondroplasia is a rare disorder occurring 1 in 15,000 to 1 in 40,000 live births. It is, however, the commonest cause of short-limbed dwarfism. It is a genetic disorder and inherited as an autosomal dominant trait but most cases (80%) are due to mutations of fibroblast growth factor receptor 3 (FGFR3) [1-3]. These individuals have normal mental and sexual development, and life span may be normal. Certain gynaecological problems like infertility, menorrhagia, dysmenorrhoea, leiomyomata and early menopause are more common in these patients. Information regarding obstetric behaviour in achondroplastic females is scarce in literature. However, problems such as pre-eclampsia, polyhydramnios, respiratory compromise, contracted pelvis necessitating lower section caesarean section, prematurity and foetal wastage, etc, have been reported[1].Early registration of pregnancy of an achondroplastic female is essential as serial ultrasonograms are required to assess any foetal influence of this disease, efficiently. Typical warning signs of the foetus developing achondroplasia are rhizomelic shortening of limbs, narrowing of the interpediculate distance of lumbosacral spine, rounded iliac wings and bilateral collar hoop sign of the proximal femurs.The confirmatory test is the heterozygote mutation in Exon 8 of the fibroblast growth factor receptor III (FGFR III). In our case we were constantly on the lookout for these features, but the developing foetus showed no telltale signs of achondroplasia.The most alarming aspect in achondroplastic mothers is an elevated mortality rate of the foetus, particularly in those who are homozygous for the achondroplastic gene. On top of all that, our patient had the simultaneous stenoses of the aortic and pulmonary valves, for which she had been symptomatic since childhood. Early attempts for surgical corrections were met with mortality. The reason for this was either incomplete diagnosis or incomplete correction of both the obstructive lesions. The first report of successful surgical repair came from Steinberg et al. in 1967. The catch was that both obstructive lesions had to be diagnosed and corrected simultaneously in same sitting. If aortic obstruction alone is relieved, an insufficient amount of blood enters the left side of heart leading to severe peripheral hypotension and death. In contrast, if the pulmonary obstruction alone is relieved, a flooding of blood into the lungs leads to pulmonary edema. With present day surgical techniques and knowledge, better results are seen. **Balloon valvuloplasty** has emerged as an attractive alternative and can be repeated over time, if required. In our patient, balloon valvuloplasty was successful in relieving pulmonary stenosis and decreasing the aortic stenosis thus obviating the need for surgery.To date, four reports of successful balloon valvuloplasty of combined AVS and PVS have been reported. Initially referred to our hospital for medical termination of pregnancy in view of the above, the credit goes to our distinguished cardiologists who alleviated the symptoms to a large extent by performing balloon aortic and pulmonary valvuloplasties; the onus was on our department to plan up her confinement and execute the delivery in a smooth and efficient manner. As we looked up for literature support we found very few articles pertaining to the successful management of pregnancies in achondroplastic females, and those addressing the management of achondroplastic pregnant women with heart disease were practically nil. Hence during the period of our patient's confinement teams of cardiologists and obstetricians were constantly on standby to observe any abnormalities that might occur. Serial 2D echoes and ultrasonograms were essential to assess any signs of cardiac decompensation or thromboembolic events and any foetal abnormalities that might occur. This case is unique as she was both achondroplastic and a symptomatic cardiac patient from early childhood and yet a concerted and well coordinated effort from the cardiologists, radiologists, anaesthetisiologists and obstetricians culminated in a successful delivery. This episode can be a reference for future cases in which obstetricians face a dilemma when faced with patients with multiple congenital defects, about whether to opt for medical termination of pregnancy or to go ahead with the gestation.Thus with a combined multidisciplinary approach involving **Obstetricians, Cardiologists, Radiologists & Anaesthesiologists,** this high risk patient was managed without any complication.

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