Abstract:
Ellis-van Creveld syndrome is a rare genetic disorder. About 300 cases have been reported worldwide. Amongst the affected population more than half do not survive beyond infancy or childhood. Thus we are presenting this case of a 28 year old short primigravida with 8 months gestation with positive family history with complaints of breathlessness and chest pain and an echocardiography revealing common atrium with severe pulmonary hypertension. Associated clinical features of short limbs, postaxial polydactyly and narrow chest led to the diagnosis of Ellis-van Creveld syndrome.

Keyword: Ellis-van Creveld, polydactyly, common atrium

Introduction:
Heart disease complicates 1% of all pregnancies. Women with congenital heart disease have a worse prognosis than those with other cardiac lesions. The prognosis depends on the degree of cyanosis and the severity of associated pulmonary hypertension rather than the actual abnormality. Our patient presented with one such congenital heart disease, a combination of single atrium with severe pulmonary hypertension in her 8th month of pregnancy. The rarity of the condition and the severity of the disorder made the situation a challenging one.

CASE REPORT
A 28 year old primigravida with 8 months amenorrhoea with EDD on 09/02/13 presented with complaints of cough, breathlessness, orthopnea and occasional chest pain. There was no history of fever, paroxysmal nocturnal dyspnoea, syncopal attacks or palpitations. She had been evaluated for breathlessness at 5 months amenorrhoea with an echocardiography which showed a single atrium. Fetal anomaly scan and fetal echo were done at the same time which showed normal study. There was history of on and off breathlessness and chest pain since childhood. She was of normal intelligence and an average student at school. Her elder sister had died at 3 years of age due to congenital
heart disease details of which are not available. She was also short for her age. Her parents had a 3rd degree consanguinous marriage.

On examination she was short statured with height of 118 cms, moderately nourished, no pallor, pedal edema, cyanosis or clubbing. She had postaxial polydactyly in all four limbs with narrow chest. Cubitus valgus was seen in both upper limbs. Pulse rate was 82/min, regular rhythm with normal volume. Jugular venous pressure was not elevated. Cardiovascular examination revealed apex beat at 4th intercostal space lateral to the midclavicular line, no parasternal heave, no suprasternal pulsations or any other visible pulsations. First heart sound was heard normally. Second heart sound was widely split with loud P2. Grade 4/6 systolic murmur was heard in the tricuspid area which increased in intensity with inspiration. Bilateral breath sounds were heard normally. No added sounds heard. Obstetrical examination revealed uterine size of 30 weeks with good fetal heart rate.

Her hemoglobin was 11.4gm%. Other routine blood investigations were normal. At room air, her oxygen saturation was 90% at which she was comfortable. Echocardiography done showed common atrium, moderate tricuspid regurgitation with severe pulmonary hypertension. Cardiology opinion was taken according to which she was started on antibiotics and frusemide 40 mg OD. Inspite of our best efforts she deteriorated during her stay in the hospital and succumbed to cardiac failure.

**DISCUSSION**

Short stature, short limbs, postaxial polydactyly and common atrium with positive family history all point to a diagnosis of Ellis-van Creveld syndrome. It is a rare autosomal recessive disorder characterised by a tetrad of chondrodysplasia, ectodermal dysplasia, postaxial polydactyly and congenital heart disease.\(^2\) It is caused by mutation in EVC and EVC2 genes which are located on short arm of chromosome 4. The exact incidence is not known but it occurs in about 1 in 60,000 newborns worldwide. Incidence is higher in old order Amish population of Lanchester county, Pennsylvania.\(^3\) Males and females are affected equally. Clinical features include short limbs, short ribs, postaxial polydactyly, dysplastic nail and teeth. Congenital heart defect, most commonly a defect of primary atrial septation producing a common atrium is seen in 60% of the affected individuals.\(^4\) Also seen in some cases are peg teeth, widely spaced teeth, natal teeth, short upper lip and scant scalp hair.\(^5\) Thoracic dysplasia and cardiac anomalies lead to death due to cardiorespiratory insufficiency in infancy in more than 50% of the patients.\(^6\)

Diagnosis is made by presence of clinical features, imaging studies and genetic testing. Imaging studies in the form of X-ray may be used to assess the degree of skeletal defects. Radiation exposure limits its use in pregnancy. Associated CNS and renal malformations if present can also be detected by MRI Brain and USG kidney. Echocardiography is done to assess cardiac...
status. Along with giving information regarding the structural defect it can also assess severity of pulmonary hypertension if present.

Molecular genetic diagnosis is by sequencing of EVC and EVC 2 gene mutations. Few research centers conduct this study due to rarity of the disease and high cost. It wasn’t done in our patient.

Prenatal diagnosis has a place in cases where there is positive family history. Amniocentesis and chorionic villus sampling confirms the diagnosis. Anomaly scan to look for features of Ellis van Creveld syndrome and fetal echo at 18 weeks of gestation also can be done.

Severe pulmonary hypertension with pregnancy as seen in our patient carries high mortality rate in the range of 30 to 50 %. The physiological changes that occur during pregnancy, labour and peripartum period are poorly tolerated in these patients with majority of deaths occur during labour and within one month postpartum period. The increase in cardiac output during labour and postpartum period due to fluid shifts may lead to sudden right heart failure. At delivery the excessive blood loss reduces preload which results in an inability to overcome high pulmonary vascular resistance. Both these situations lead to reduced ventricular output, myocardial ischaemia, arrhythmias, ventricular failure and sudden death. Pregnancy is thus best avoided in such cases.

If pregnancy does occur, termination is offered because of maternal risks. If she is well into pregnancy at the time of diagnosis and pregnancy is continued a multidisciplinary team of obstetrician, cardiologist and critical care specialist must work together. The risk of mortality is comparable with both vaginal delivery and cesarean section. Intrapartum inhaled nitric oxide or intravenous prostacyclines have been described as they cause vascular dilatation thus lowering pulmonary vascular resistance and providing better oxygenation although their efficacy is better proved in primary pulmonary hypertension.

Although recent advances in management of pulmonary hypertension in pregnancy are reducing the mortality rates, it is advisable to discourage pregnancy in these patients. Also this shows a need of increased awareness of such conditions in general population and place of genetic counseling in people with family history of such disorders.

References:

2 Arya L, Mendriatta V, Sharma RC, Solanki RS, Ellis-van creveld syndrome.


5 About.com Ellis-van Creveld Syndrome Harold Chen M.D, Ellis-van Creveld syndrome, emedicine.medscape, August 2011

6 Natural standard.com; Ellis-van Creveld syndrome.


