Three siblings with the extremely rare geleophysic dysplasia and different severity of cardiac involvement.

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Background:
Geleophysical dysplasia (GD) is an extremely rare autosomal recessive skeletal dysplasia resembling lysosomal storage disease, characterized by short stature and short limbs, joint contracture and cardiac involvement. The most frequent cardiac lesions described being the mitral valve, the aortic valve followed by pulmonary valve and the least is tricuspid valve. It has been described in few cases worldwide.

AIM: To study the progression of cardiac lesions in patients with Geleophysical dysplasia.

Methods and Results:
We reviewed the cardiac lesions in three female siblings of a Saudi family who showed different severity of the valvular involvement. The three sisters are seven, six and two years old. They were born to first cousins parents who also have 3 years healthy son. They had history of respiratory problems requiring frequent hospital admissions. For last 3 years weight and height grew parallel but considerably below 3rd centile. Furthermore they shared dysmorphic features of small and broad hands and feet, hypertelorism, depressed nasal bridge and anteverted narse. They have limitation of flexion of their hands with bilateral contractures of both elbow and knee joints. They have happy and friendly personality with normal intelligence. Genetic studies confirmed the heterogenicity of the ADAMTSL2 gene.

All sisters have associated cardiac lesions with different severity. The eldest has mild pulmonary and aortic valve stenosis. The middle sister has thickening of mitral valve leaflets without stenosis. The youngest sibling has severe aortic valve stenosis and mild pulmonary valve stenosis.

We observed that the valvular involvement remained static in first two patients over period of 3 years. This observation helped to manage the third case in a more conservative way despite severe aortic stenosis and after one year follow up the aortic valve gradient remained the same.

Conclusions:
In geleophysic dysplasia cardiac valves are usually thickened leading to stenosis. Most of the reports suggested progression of the disease. In our three cases the youngest has the most severe form of valve involvement. This suggests that the expression of the gene could determine the severity of the disease, rather than the belief of progression with time.