CASE REPORT

Familial Heart Disease with Skeletal Malformations- Holt – Oram Syndrome

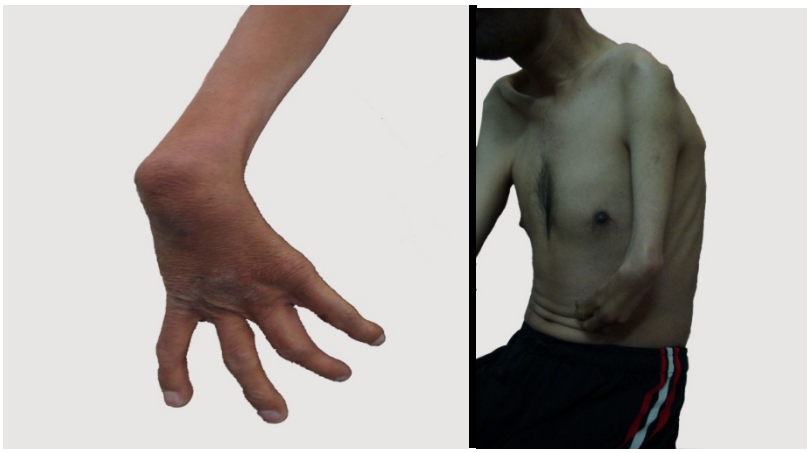
Karim Al-naffi

It is well known that congenital heart disease and skeletal deformities occur together in several clinical syndromes such as 13-15 or DI trisomy,16-18 or EI triosomy, mongolism, XO syndrome, XXXXX, XXXY, Ellis Van Crevald syndrome and Marfan syndrome yet there are a lot of skeletal deformities which is associated with congenital heart disease ,so if found such deformities we should examine the heart of these patients for congenital anomalies..
Below is a case repot of a family having one of these condition namely Holt Oram Syndrome, with review of this hereditary disease, and a list of her diseases which have skeletal anomalies and congenital heart disease

Case report

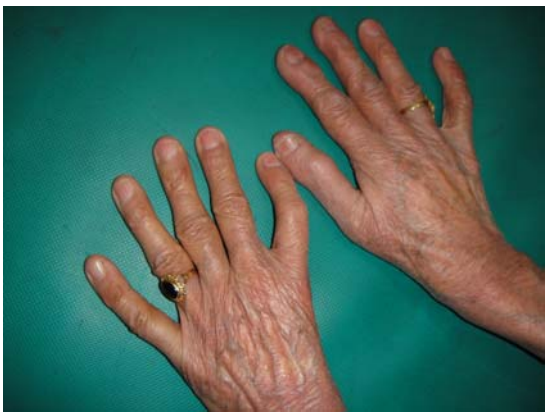
Q R is 27 yr old patient presented with cough & dyspnea after upper respiratory tract infection. He is a member of a family of five brothers & one sister, his father is the son of his mother aunt O/E.
He is orthopoenic, not cyanosed, B.P100/60. Respiratory rate 30/min. JVP 5cm above costal margin
He has **skeletal deformities** which include; small atrophied left upper limb with the absence of the arm with the hand attached to the forearm, he hand which is small, the fingers shows parallel fingers including the thumb which lost apposition to the other finger in addition, the syndactly of the fingers. As seen in picture number 1., the right upper limb also is deformed with loss of the thumb, the chest is asymmetrical the right one is more promineAnt.

His mother has also deformities in her hands only, where the there is loss of the thumb apposition, she has ASD as well. One of his brothers have hand anomalies (loss of apposition of the thumb with mitral valve prolapsed), the other member of the family are normal



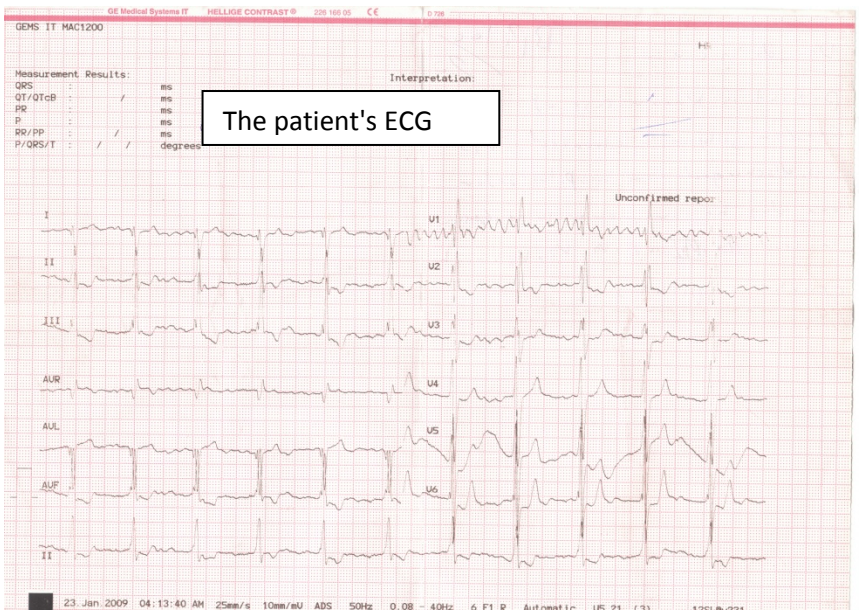
Upper limbs and shoulders of the patients

Hand of the patient



The hands of the patient's mother

Heart examination revealed apex beat in the sixth intercostals space ,with visible pulsation at the pulmonary area, there is fixed splitted second heart sound in the pulmonary area with ejection systolic murmur grade 3,Also there is long systolic murmur grade 2-3 at mitral area with radiation to the axilla. **Echocardiography** shows big ASD (picture) with left to right flow, dilated both ventricles with diminished contractility, mitral valve prolapse, in addition, there was tricuspid regurgitation.



His ECG shows right axis deviation, partial RBBB&RVH.