CASE REPORT

Progeria

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BACKGROUND:
Hutchinson-Gilford Progeria Syndrome (HGPS) is a very rare genetic disorder with a frequency of 1 in 8 million live births. It is characterized by premature aging phenotype. The median age at death is 13.4 years.
KEYWORDS: hutchinson-gilford progeria syndrome, failure to thrive, alopecia .

INTRODUCTION:
Progeria is of considerable interest because of its striking features, which resemble accelerated aging. this rare syndrome has a reported incidence of approximately 1/8 million. As result of sever failure to thrive, affected children do not become sexually mature and reproduce. parental age is significantly increased but there is no increase in consanguinity. Therefore each child with progeria most likely represent a new sporadic dominant mutation [(1)]. Mental development is not affected[(2)]. The development of symptoms is comparable to aging at a rate six to eight times faster than normal, although certain age-related conditions do not occur. Specifically, patients show no neurodegeneration or cancer predisposition[(3)].No treatments have been proven effective[(4)]. Most treatment focuses on reducing complications (such as cardiovascular disease) with heart bypass surgery or low-dose aspirin[(5)]. Growth has been attempted[(6)].There is no known cure. Few people with progeria exceed 13 years of age[(7)] At least 90% of patients die from complications of atherosclerosis, such as heart attacks or strokes[(8)].Scientists are particularly interested in progeria because it might reveal clues about the normal process of aging[(9)].

CASE REPORT :
6-years-old girl child presented with progressive history of coarsening of skin, failure to thrive and inability to squat for the past three to four years. The child had also developed global alopecia over the past few years. The perinatal history was uneventful. She was apparently normal till one year of age when the parents started noticing their child fail to grow in spite of normal intelligence. they began to attend many hospitals and private clinics and investigations were done to exclude any medical illnesses that lead to failure to thrief e.g. thyroid function test, growth hormone assay in addition to general investigation like complete blood picture ,general urine and stool examination all investigations were normal she received a lot of drugs in form of antibiotics because of suspicion of urinary tract infections and multiple tonics drugs but without benefit also the parents notice that the appearance of their child is different from her three brother(two female and male).father and mother are second degree relative. No family history of similar complaints could be elicited. And she was fully vaccinated .On examination child to be of short stature and malnourished her body weight was 11 Kg which is bellow 3rd percentile also her height was 90 Cm which is bellow 3rd percentile[Figure-1-] . Eyes appeared prominent with hypo plastic chin. Multiple patches of coarse and thickened skin, especially over the dorsum of the hands and shoulders. The terminal ends of the fingers appeared broad and stubby. her hair show male pattern baldness, bossing of forehead and dilated scalp veins, face was small with senile appearance,mandibule was hypo plastic [Figure-2-] and teeth was abnormal and crowded , skin thinning with areas of hyper pigmentation with scleroderma, voice was high pitch which is evidence of laryngeal atrophy . Based on the history and clinical findings a provisional diagnosis of progeria was made. Radiological investigations show evidence of ostiopenia[Figure-3-Aand-B] and bone age was compatible to chronological age.ECHO-study and ECG both were normal ,also we send her for serological tests to exclude celiac disease which was normal.
Biochemical investigations were normal except for increased atherogenic index [Table-1-] which indicate evidence of atherosclerosis. The clinical finding ,radiological findings and biochemical results confirmed the clinical diagnosis of progeria

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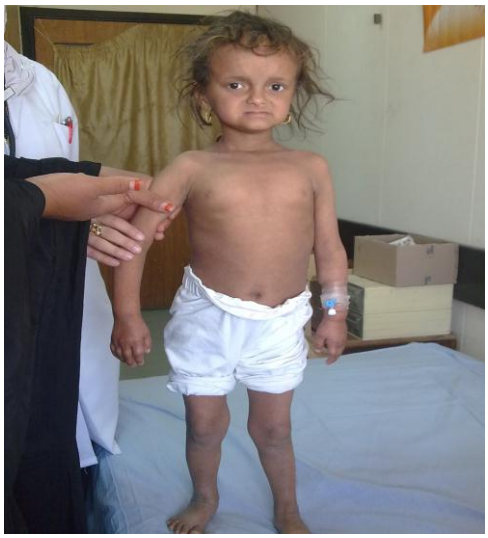

Figure 1:Short stature with senile appearance

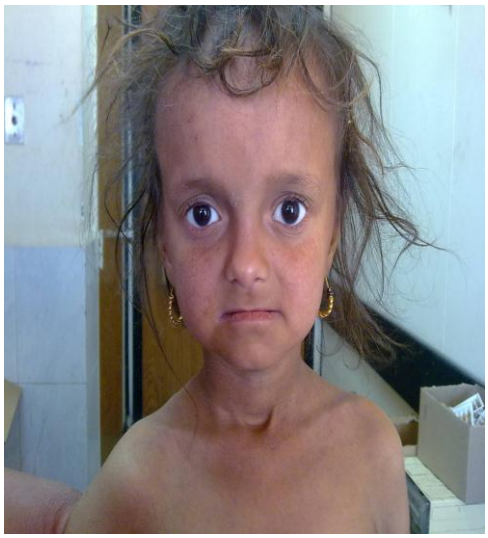

Figure 2:Bossing of forehead ,prominent eyes,hypoplastic chin ,male pattern baldness, dilated scalp veins and senile appearance.

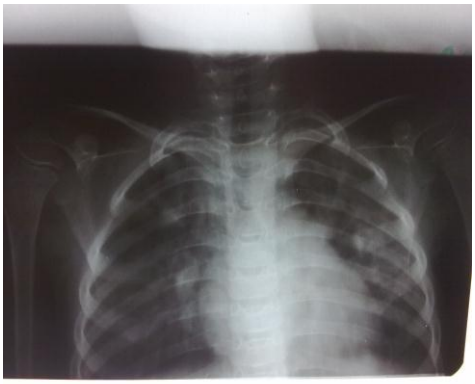

Figure 3: A- osteopenia of clavicle.

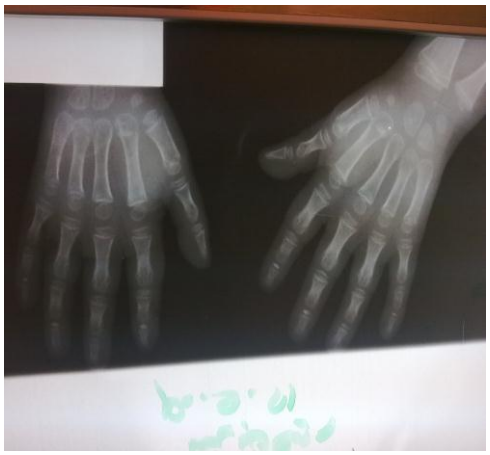

Figure 3: B-bone age is 6 years with osteopenia.

Table1 :Lipid profile value of the patient.

Test	Value mmol/l	Normal values mmol/l
S.Cholesterol	5.9	3.6-6.5
S.Triglycerides	1.1	0.9-2.4
S.HDL	6.5	0.9-1.4
S.LDL	4.9	1.8-4.3
S.VLDL	0.5	<0.53
Ahterogenic index	9.8	Less than 4.5

DISCUSSION:

Progeria was first described in 1886 by Jonathan Hutchinson and also described independently in 1897 by Hastings Gilford. The condition was later named Hutchinson-Gilford Progeria syndrome (HGPS)[10]. Is an extremely rare genetic condition where symptoms resembling aspects of aging are manifested at an early age. About 1 in 8 million babies are born with this condition, and most affected children usually die at around age 13 years[11]. The earliest symptoms include failure to thrive and a localized scleroderma like skin condition[12]which is similar to earliest symptoms in our patient . As the child ages past infancy, additional conditions become apparent. Limited growth, alopecia, and a distinctive appearance (small face and jaw, pinched nose) are all characteristic of progeria. The people diagnosed with this disease usually have small, fragile bodies, like those of elderly people. Later, the condition causes wrinkled skin, atherosclerosis, and cardiovascular problems[8] All of these symptoms occures and becomes obvious in our case but there is no clinical evidence of cardiovascular problems also the ECG recording and ECHO study were normal in spite of biochemical changes indicating premature atherosclerosis which can be explained by short duration of illness and there is possibility to develop cardiovascular problems later on ,so I put my patient on aspirin therapy in order to reduce or minimize the cardiovascular complications .mental development is not affected [2]. As in our case. As the syndrome is very rare so there are few cases reported in the world like ,study from the Nether land has shown an incidence of 1 in 4 million birth [13].A family from India had five Progeria children, two of which are now deceased. [14]. Also One of the authors, Leslie Gordon, was a physician who didn't know anything about progeria until her own son, Sam, was diagnosed at 21 months[15].

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