ABSTRACT
Background: Infantile systemic hyalinosis (ISH) is a rare autosomal recessive disease that happens within the
first weeks of life. It is present in Saudi Arabia due to positive parental consanguinity. A suggestive diagnostic
.criterion was proposed which would be helpful in determining ISH
Case Presentation: Seven children were diagnosed with ISH based on clinical pictures and histopathological
examination. A prospective consecutive and observational study was designed to determine -the diagnostic cri
teria of ISH. On clinical examination, all patients had joint contractures which led to frog-like -position, hyper
pigmentation overlying the distal joints, in addition to failure to thrive. Only five patients of (seven (71.42%
had erythematous papules coalesce to form plaque on the posterior neck. Out of seven, four (patients (57.14%
had nodules (perianal and subcutaneous), and gingival hypertrophy. Three patients of seven (42.85%) had
intractable diarrhea. Just one patient (14.28%) had small pearly skin papules on the face. Hyaline deposits
were evident in histopathological examination of the skin biopsies for all the patients. Although, the severity
index and genetic study were unavailable
Conclusion: ISH is a congenital disease characterized by excessive hyaline deposits. Diagnostic criterion was
evaluated for the seven patients as three major criteria and five minor criteria. It was suggested that the
presence of all three major criteria or two major and two minor criteria is essential for the .diagnosis of ISH
The diagnostic criterion would help in the early evaluation of the disease prognosis and would enhance early
.intervention
.Keywords: Infantile systemic hyalinosis, autosomal recessive, joint contracture