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J Clin Exp Cardiolog 2018, Volume 9
DOI: 10.4172/2155-9880-C11-120

Ellis van creveld syndrome: A rare case report

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Ellis Van Creveld Syndrome is a rare genetic disorder having autosomal recessive inheritance, mostly affecting the Amish population of Lancaster county in Pennsylvania in the US, with prevalence rate of 1/5000 at live birth. In non-Amish population, the birth prevalence is 7/1,000,000. Classical manifestations of the disease include small stature, short limbs, hypoplastic finger nails, bilateral post axial polydactyly, hypoplasia of the enamel, hypodontia and malocclusion. Heart defects, especially abnormalities of atrial septation has been reported in 60% of the cases. Very few cases of Ellis Van Creveld Syndrome have been reported in Pakistan. We hereby present a case of 13-year-old male child presenting with classical oral, chondroectodermal and cardiac manifestations of this syndrome.

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