



pairs and in 2004 the complete human DNA sequence was published. In addition they noted that many genes can have multiple functions. ability to identify the gene that is responsible of an inherited single gene disorder and its diagnostic application will allow better understanding of diseases pathogenesis as well as possible therapeutic applications. This could be achieved through the identification of the human disease candidate gene by the use of animal models of disease or by homology. New disease genes can now be identified using genetic databases [8,9]. During 1980s recombinant DNA techniques allowed mapping through positional cloning which lead to purely positional cloning of the gene by its location without knowing its function [10]. Sequencing technology developed to Exome sequencing which is the

lip and or palate could be part of a syndrome as a result of single gene disorder or in few situations, as a result of multigene abnormalities. Lip and or palate also might be an isolated defect which is more common. Possible genes related to nonsyndromic are: Blood clotting factor XIII gene (*F13A*), *endothelin-1* gene and other involved genes. An example of syndromic is Di George Syndrome where its pathogenesis is attributed to failure of neural crest cells to migrate in the areas of 3<sup>rd</sup> and 4<sup>th</sup> brachial arches. It is related to the deletion in the chromosome number 22 and mutation in *TBX22* gene [30,31]. Another example is holoprosencephaly syndrome which is caused by mutation in *SHH*

Osteitis deformans or can be called Paget disease of bone is a form of osseous dysplasia with increased bone turnover in all body bones [20,24,57]. Its genetic mechanism is through a gene called Sequestosome 1 gene (*SQSTM1*) which is a  $\kappa$  protein in the NF kappaB pathway resulting in inactivation mutations in TNFRSF11B that encodes osteoprotegerin [13,16,52,58-60].

Cherubism is an autosomal dominant inherited disease [61,62].



requires muscle biopsy to test the reaction with the anesthesia.  
most likely cause is mutation in ryanodine receptor gene (*RYR1*).

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