WILEY'S

International Journal of Dentistry and Oral Health

Volume 8 Issue 2,
March 2022

Citation
Ohoud Tawfig Zain Elabdeen. (2022). Case Report: Digeorge Syndrome (22q11.2 Deletion Syndrome) Int J Dent & Ora Hea. 8:2, 7-13

ISSN 2471-657X

Published by
Biocore Group |
www.biocoreopen.org/ijdoh/archive.php

Case Report

Case Report: Digeorge Syndrome (22q11.2 Deletion Syndrome)

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Article History: **Received:** March 11, 2022;
Accepted: March 18, 2022;
Published: March 23, 2022.

Abstract
Digeorge syndrome is also known as velo-cardio-facial syndrome, it is a complex genetic multi-organ disorder occurs due to deletion in 22q11.2 chromosome. Patients usually presented with congenital heart defects, characteristic faces that includes: retrognathisa or micrognathia, low set ears, hypertelorisim. They may also present with ophthalmic, renal, pulmonary and gastrointestinal abnormalities. We report a case of 11 years old male with this type of syndrome and his oral condition.

Keywords :
Digeorge Syndrome, Chromosomal Deletion Syndrome, Multiorgan Disorder and Skin Abnormalities

Conflicts of interest:
I declare no conflict of interest.

Consent:
Written and verbal consent was taken from the patient mother prior to taking pictures and submission of this report.

Introduction:
Digeorge syndrome is a chromosomal defect disorder that occurs due to deletion of chromosome 22q11.2, it has varying degrees of severity with incidence of 1 in 4000 live births[1] , the clinical problems of this syndrome usually comprise congenital heart defects, hypoplasia of the thymus, some dysmorphic facial features such as: long face, low set ears, hypertelorism, retrognathia or micrognathia short philtrum and thin lips.Some autoimmune disorders may also appear as T-cell dysfunction , renal and gastrointestinal , pulmonary as well as ophthalmic abnormalities are also reported[1,6].

Children with such syndrome may present with some behavioral, speech, communication and psychiatric disorders for example: attention deficit hyperactivity disorder (ADHD) and learning difficulties, also in adults some psychotic disorder is also reported as: schizophrenia[3].

In addition some patients presented with severe chronic arthritis, few researches reported increased prevalence of chronic arthritis in association with IgA deficiency, but the reasons for this association are unknown[2]. Arthritic joint exhibits cardinal signs of pain, swelling, heat and may be loss of function [3]. Sometimes high fever, skin rash, serositis and uveitis may develop as extra articular manifestation[4].

There is only few reports on patients with digeorge syndrome presenting with skin rash or erythroderma and eczema [4].

There is limited reports on the dental and oral manifestations of patients with digeorge syndrome some noted teeth hypoplasia and enamel opacities as well as tooth agenesis and supernumerary teeth [5]

Case Report:

An 11 years old Sudanese boy came to our clinic complaining of pain on the floor of the mouth. Upon taking his medical history mother reported that his is diagnosed with Digeorge syndrome and he suffered from congenital heart disease, vomiting and indigestion which was one of this syndrome abnormalities and he was treated accordingly. Also he had an eye operation when he was 6 years old because he started to lose his sight, the opthalmologist report reviled that the patient have corenal leukoma in both eyes, photophopia and neovascularation of the left eye .

On eye examination he still photophopic but he can see fine with the right eye, the left one regained only half of it's function.

Patient is suffering from some behavioral and communication problems with some learning difficulties so it was a challenge to connect with him, he might need referral to a specialist regarding this issue.

On the extra-oral examination :patients have some dysmorphic facial features such as: long face, low set ears, hypertelorism, micrognathia short philtrum and thin lips, his left eye showed corneal leukoma.

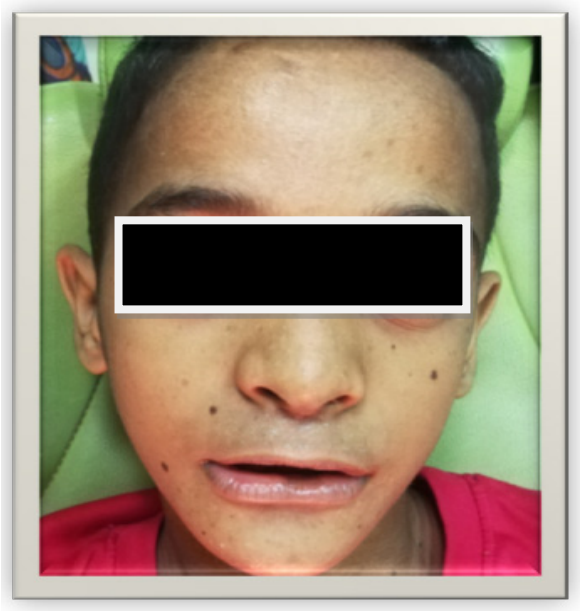


Figure (1) : shows some the dysmorphic facial features.

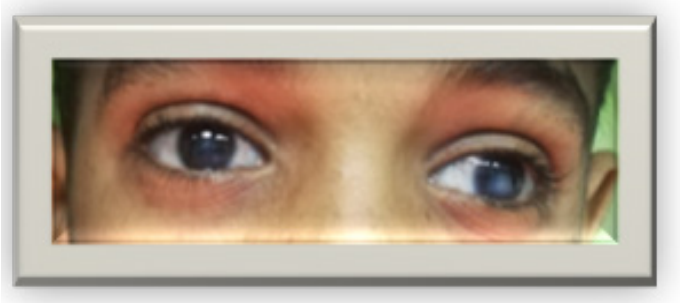


Figure (2): shows left eye leukoma and hypertelorisim.

The patient also presented with sever skin dryness and rash (eczema and scaly skin) as well as swelling of the joints that he suffered from since 2 years old. He is using some corticosteroids and body moisturizers to keep the skin moist and prevent it from scaling.

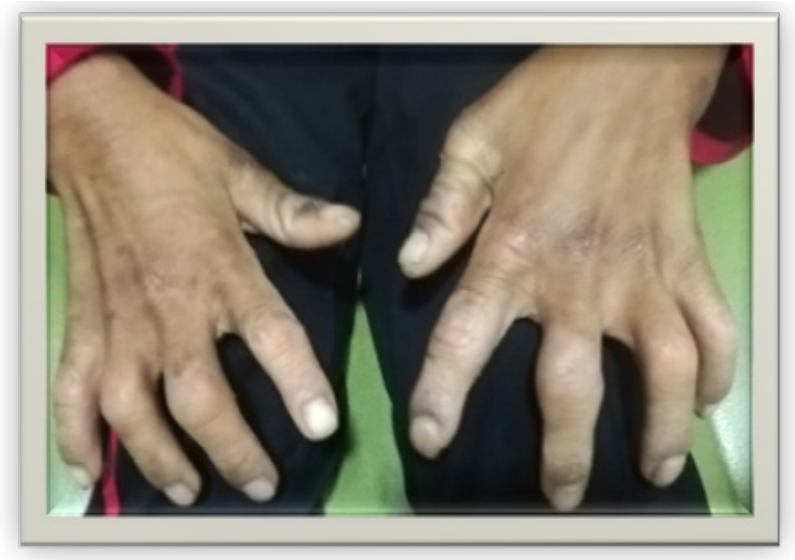


Figure (3) : shows arthritis in both hands and fingers joints disfigurement.

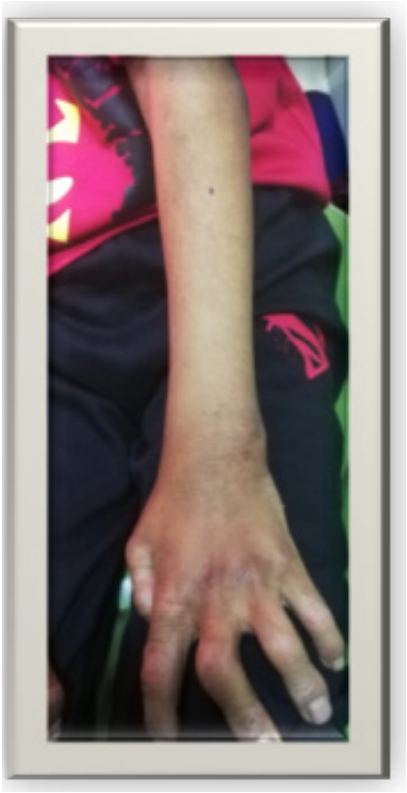


Figure (4): shows skin rash

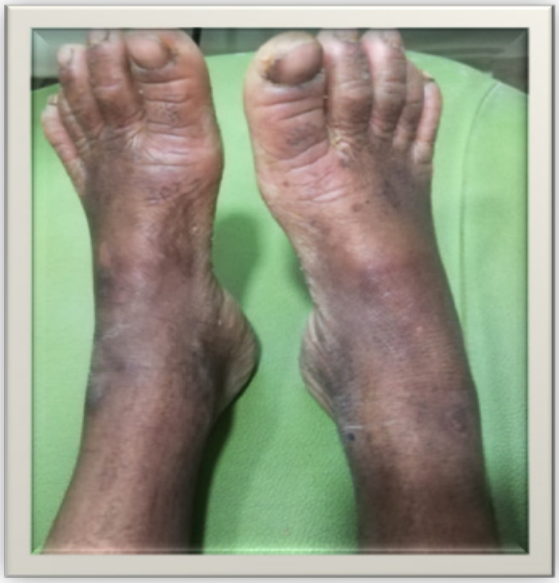


Figure (5): shows skin rash on the feet and scaly skin

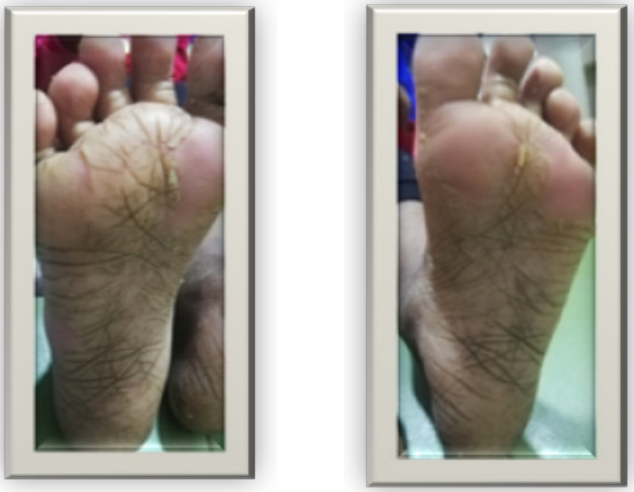


Figure (6): shows very dry and scaly feet soles

On examination of the intra-oral cavity the pain was from an ulcer located on the left side of the ventral surface of the tongue due to sharp tip of the lower left canine. Smoothness of the tip of both canines was done and prescription of oral gel containing hyaluronic acid was given.

Also his gingiva was inflamed and gross blaque accumulation was found so scaling is done and mouth wash was given as well. In addition to that some unerupted teeth were found (delayed eruption) so radiograph was taken (OPG) to see if they exist.

Deep bite was also noted.

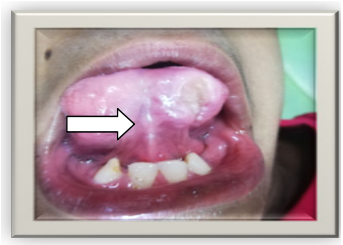


Figure (7): shows traumatic ulcer on the left side of the ventral surface of the tongue.

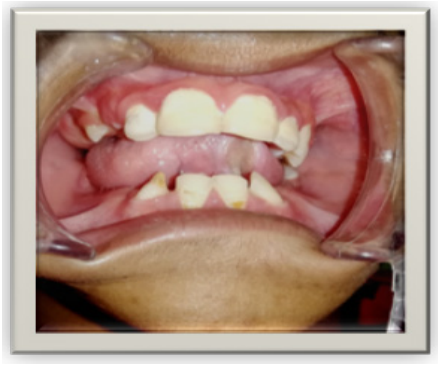


Figure (8): shows gross blaque accumulation and inflamed gingival as well as some unerupted teeth.

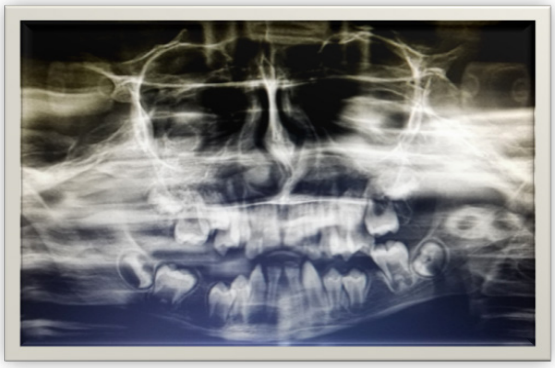


Figure (9): shows the OPG of the patient and the presence of unerupted teeth. Also spaces between the mandibular second pre- molars and the first molars are detected and the co-patient was asked if the child had any previous extractions and she said he did not have any dental treatment before.

After 6 month the patient came for follow up and he was complaining of the same ulceration but it was on the right side of the ventral surface of the tongue due to the eruption of the mandibular first pre-molar and smoothening of it's tip was also done using low speed handpiece and same medication was prescribed.

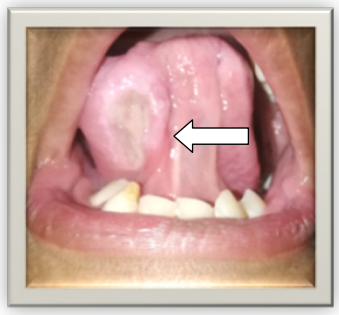


Figure (10): shows traumatic ulcer on the right side of the ventral surface of the tongue.

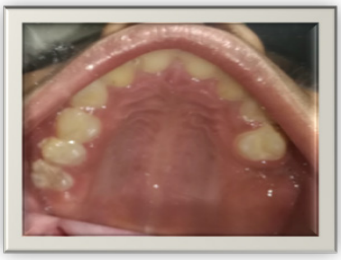


Figure (11): shows the eruption of the maxillary left first molar and the right is soon to erupt.

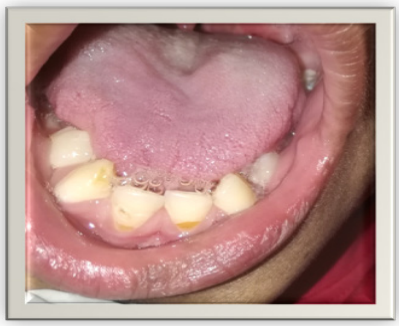


Figure (12): shows the lower jaw with both first premolars erupted. The lower right 1st premolar tip is sharp causing the trauma.

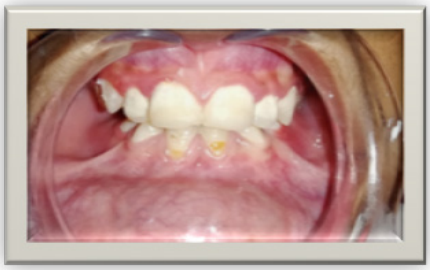
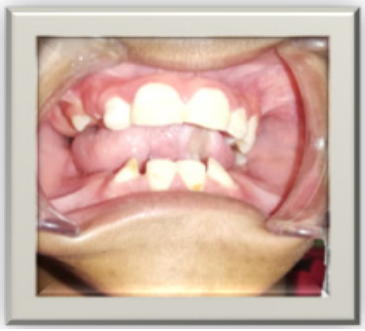
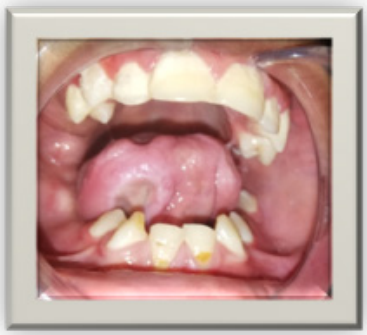


Figure (13): shows inflamed gingival especially in maxillary teeth and deep bite.

(1)





(2)

Figure (14): (1): shows the first time the patient came to the clinic.
(2): shows the eruption of the delayed teeth after six month plus the trauma from the right 1st premolar.

Discussion:

Several abnormalities were described in Digeorge syndrome but in this case our patient suffered from only few manifestations as the dysmorphic facial structures , congenital heart defects, some gastrointestinal problems and finally some skin problems and rheumatoid arthritis. Also he suffers from some learning difficulties and behavioral issues.

To date only few cases of Digeorge syndrome suffered from eczema[4] but it is found in this case with the rheumatoid arthritis hence it is rare as well. Specific medications should be prescribed to manage these conditions so the mother was advised to contact the patient pediatrician

Regarding the oral and dental presentations of this syndrome there is no enough evidence [6] to generalize the presentations that our patient came with that include: delayed eruption of permanent teeth, gingival inflammation and some teeth hyopoplasia with sharp edges that may cause ulceration.

In conclusion this case report may create awareness among Sudanese dentists if they came across such a syndrome what to expect and how to manage them. Further reports and researches regarding the dental presentations and abnormalities in such patients should be done.

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