Noonan Syndrome: a case report

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Abstract

Noonan syndrome is a genetic multisystem disorder that causes abnormal development of the body. It is characterized by distinctive facial features, short statue, webbed neck, developmental delay, learning difficulties, congenital heart defects, and increased bleeding tendency. Noonan syndrome is caused by a genetic mutation, commonly in the Ras/MAPK (mitogen-activated protein kinase) cell signaling pathway, which is essential in the regulation of cell cycle, differentiation, growth and cell senescence. The mutation can be inherited in autosomal dominant pattern or often de novo mutation. Mutation of genes encoding proteins in Ras/MAPK pathway affects growth and development of a patient with Noonan syndrome.

This presentation discusses a 16-year-old female patient who was referred to Tufts School of Dental Medicine pediatric clinic due to multiple caries.
The patient's medical history is significant for Noonan syndrome, ADHD, exercise-induced asthma, and history of cardiac surgery when patient was one year old. Current medication is albuterol as a rescue inhaler for asthmatic attack. Patient did not report any drug allergies. Extra oral clinical examination revealed short stature, wide-set and down slanting eyes and short neck. Intra oral examination revealed high arched palate, malocclusion, micrognathia, multiple caries, anterior open bite, over retained primary teeth and unerupted #6 and 11. This report includes clinical findings associated with Noonan syndrome and dental management of patient with Noonan syndrome.

Background

- Noonan syndrome is a dominantly inherited genetic multisystem disorder that is affecting 1:1000 to 1:2500 live birth[1,2,4]
- Affected individuals have abnormal development of the body that is characterized by distinctive facial features, short statue, broad, webbed neck, developmental delay, learning difficulties, congenital heart defects, and increased bleeding tendency[1-4]
- Genetic mutation in Ras/MAPK (mitogen-activated protein kinase) cell signaling pathway that regulation of cell cycle, differentiation, growth and cell senescence affects growth and development of a patient with Noonan syndrome[2,3,5]

Clinical Presentation

A 16-year-old female patient was referred to Tufts School of Dental Medicine Pediatric department due to multiple caries. The patient's medical history is significant for Noonan syndrome, ADHD, exercise-induced asthma, and history of cardiac surgery when patient was one year old. Current medication is albuterol for asthma. Patient did not report any drug allergies. Extra oral clinical examination revealed short stature, wide-set and down slanting eyes and short neck.(Fig. 1,2) Intra oral examination revealed high arched palate, malocclusion, micrognathia, multiple caries, anterior open bite, over retained primary teeth and unerupted #6 and 11. #11 is horizontally impacted.(Fig. 3-10).

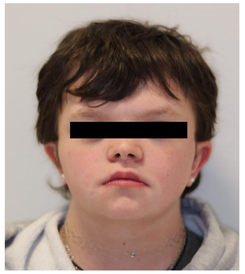

Fig.1 Facial front picture

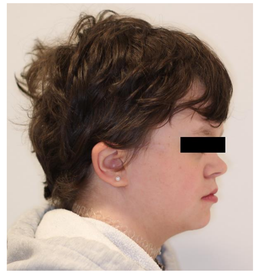

Fig.2 Facial profile

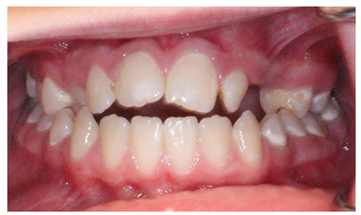

Fig.3 Intraoral center photograph

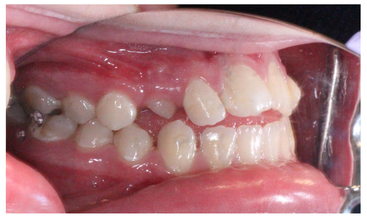

Fig.4 Intraoral right photograph

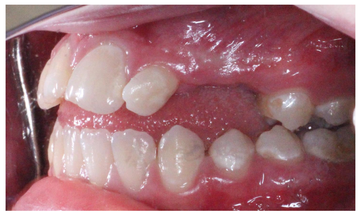

Fig.5 Intraoral left photograph

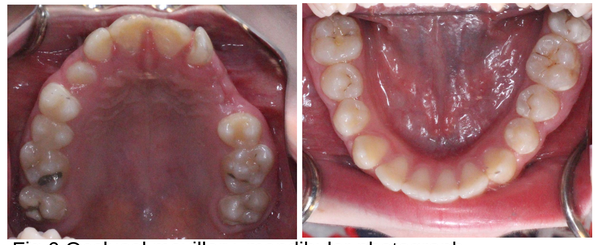

Fig.6 Occlusal maxillary, mandibular photograph

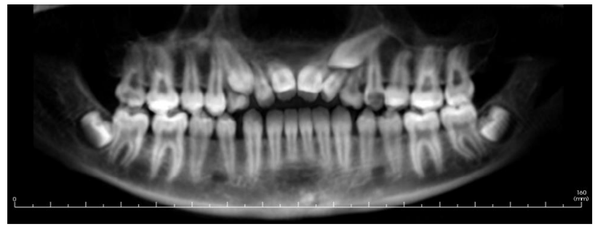

Fig.7 CBCT Panoramic

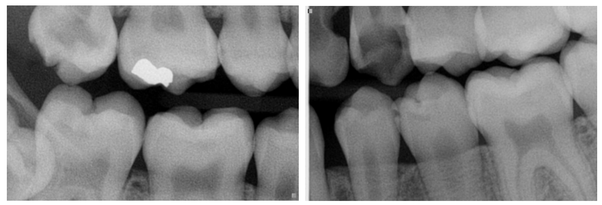

Fig.8 Bitewing radiographs

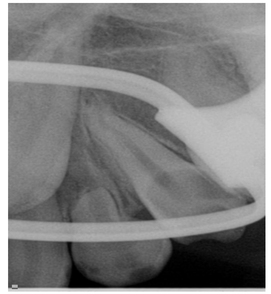

Fig.9 Periapical radiograph of impacted #11. #H, #12

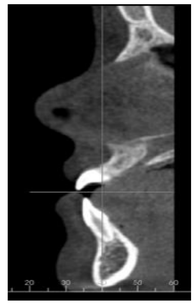

Fig.10 CBCT Anterior

Management

1. Limited Exam phase
a) Patient had multiple large carious lesions
b) Extractions of #C, #H, #12.were already done in OMFS

2. Comprehensive Exam
Treatment plan phase
a) Caries control with restorations
b) Orthodontic consult – surgical exposure #11. Comprehensive ortho treatment
c) Prosthodontic consult – Possible #2 prophylactic RCT #2. Permanent SSC on #2

3. Caries control
Treatment phase
Resin restorations with indirect pulp caps on #4(MOD), 5(DO), 7(MFL), 8(MDFL), 9(MDFL), 10(DF), 14(OL), 15(OL), 18(O), 19(DO), 20(MOD), 21(O), 28(O), 29(O), 30(MO), 31(O) were completed.

4. Maintenance phase
Due to patient's high caries risk, recommended periodic exam, prophylaxis, and fluoride varnish application every 3 months.

Conclusion

Noonan syndrome is one of the common genetic disorders that pediatric dentists may encounter. Patients could present mild characteristics and be overlooked. Early genetic counseling is recommended since each patient presents different symptoms and requires individually coordinated treatment. Recognizing patient's cardiac defects, limited growth, delayed development and learning ability, and increased bleeding tendency is important to provide safe dental management.
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References

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