



## Orofacial Aspects of Young Patients with Achondroplasia

Ambarkova Vesna\*

Department of Paediatric and Preventive Dentistry, University Ss. Cyril and Methodius, Skopje, Republic of Macedonia

### Introduction

The term achondroplasia means syndrome whose main characteristics are low growth, dwarfism, due to short limbs. Moreover, syndrome is followed by increased head, saddle nose, short palms, lordosis of lumbar spine and bulging butt and belly. Syndrome shows a high degree of new mutations. In 80% of cases of the disease are due to new point mutations and in 20% of cases can be determined through family transmission as autosomal dominant status [1]. Sick people are heterozygous for the gene that carries achondroplasia. Also homozygous cases can be met when both parents have achondroplasia. Such persons are with much more pronounced abnormalities compared to heterozygotes. Craniofacial features. The head is enlarged with prominent forehead and saddle nose. Changes are not visible at birth and occur later. Often, encountered hypoplasia of the middle third of the face and mandibular prognathism, the front teeth are crowded with malocclusions of Angle third class.

**Genetics:** Syndrome is inherited autosomal dominant. Most cases are new point mutations. It is estimated that the frequency of the gene for achondroplasia ranges from 0.00004 to 0.00014 in different populations [1]. Patients can be found on the streets, in the paintings of old masters such as Velazquez (Museum "Prado" – Madrid, Spain) in circuses, in music videos and movies.

**Frequency of occurrence:** Newborns are 1 in 10,000. Equally affected are men and women of all races.

**Etiology:** Unknown. Achondroplasia is a skeletal dysplasia characterized by disruption of endochondral ossification, primarily in the proximal thighs and upper arms, resulting in shortening of these bones and their expansion. Centers responsible for the growth of long bones are disrupted, and reduced bone growth in length, but increased their thickness. Patient with achondroplasia also very often have anomalies of other organs and organ systems, such as congenital heart defects, inflammation of the middle ear (otitis media), neurologic complications, and obesity.

**Distinctive look:** Babies at birth are usually only 46 cm to 48 cm long, dwarfism with very short legs and arms, expressed buttocks, twisted "O" feet, wobbly walking like duck. There is a delay in the motor development of the child (posture of the head, sitting, crawl, walking). Intelligence and cognitive characteristics are normal. The maximum height that reaches is up to 120 cm to 130 cm.

**Skull:** Frontal bone is significantly expressed, the root of the nose is very depressed and the nose is broad, undeveloped middle third of the face.

**Intraoral findings:** Upper jaw is underdeveloped, pseudoprognathism, extreme narrowness and overcrowding upper dental row. Kale "et al." [2] in their case report present a rare case of oligodontia in a young, female, achondroplastic patient. In their case report, Karpagam "et al." [3] present a female with severe maxillary retrognathism and vertical excess along with anterior open bite. The treatment plan consisted of modified anterior maxillary osteotomy for simultaneous vertical and sagittal augmentation along with orthodontic intervention.

**Therapy:** Application of growth hormone (Somatotropin) until puberty is obligatory, in order to reach a satisfactory height of body. Orthopedic treatments are often required, due to anomalies of the skeletal system. Sometimes it makes orthopedic extension of the upper and lower limbs to 20 cm [4].

### Orthodontic Treatment

The therapy aims to stimulate the growth of the upper jaw. In orthodontic treatment are used mobile orthodontic appliances "Y", mask Delairer (Figure 1), and fixed orthodontic appliances for the permanent dentition. In certain phases of orthodontic therapy intermaxillary rubbers are

### OPEN ACCESS

**\*Correspondence:**

Ambarkova Vesna, Department of Paediatric and Preventive Dentistry, University St. Cyril and Methodius, Mother Theresa 17 University Dental Clinic Center Sv Pantelejmon, Skopje 1000, Republic of Macedonia, Tel: +38970686333;

E-mail: vesna.ambarkova@gmail.com

**Received Date:** 03 Apr 2017

**Accepted Date:** 07 Apr 2017

**Published Date:** 12 Apr 2017

**Citation:**

Vesna A. Orofacial Aspects of Young Patients with Achondroplasia. Clin Case Rep Int. 2017; 1: 1001.

**Copyright** © 2017 Vesna A. This is an open access article distributed under the Creative Commons Attribution License, which permits unrestricted use, distribution, and reproduction in any medium, provided the original work is properly cited.



**Figure 1:** Delairer mask.

used to regulate bite. Al-Saleem and Al-Jobair [5] in their case report observed intraorally, macroglossia, tongue-thrust swallowing pattern, generalized gingivitis, posterior crossbite, anterior open bite and anterior reversed jet.

## Conclusion

In conclusion, the features of achondroplasia may lead to respiratory, neurological, skeletal, orthodontic and psychosocial problems. Dentists treating these children should be able to conduct good and appropriate dental management.

## References

1. Cohen MM. Dysmorphic syndromes with craniofacial manifestations. In: Stewart RE, Prescott GH, editors. Oral facial genetics. St. Louis: CV Mosby; 1976. p. 500-662.
2. Kale L, Khambete N, Sodhi S, Kumar R. Achondroplasia with oligodontia: Report of a rare case. *J Oral Maxillofac Pathol.* 2013;17(3):451-4.
3. Karpagam S, Rabin K, George M, Santhosh K. Correction of anterior open bite in a case of achondroplasia. *Indian J Dent Res.* 2005;16(4):159-66.
4. Rohilla S, Kaushik A, Vinod VC, Tanwar R, Kumar M. Orofacial manifestations of achondroplasia. *EXCLI J.* 2012;11:538-42.
5. Al-Saleem A, Al-Jobair A. Achondroplasia: Craniofacial manifestations and considerations in dental management. *Saudi Dent J.* 2010;22:195-9.