Studia Universitatis "Vasile Goldiş", Seria Ştiinţele Vieţii
Vol. 23, issue 3, 2013, pp. 309-311

LIVING WITH ACHONDROPLASIA – SOME MEDICAL AND SOCIAL ASPECTS

Simona DUMITRA[1*], Sabina MORGOVAN[2], Andreea DUPTA[3], Otilia MARGINEAN[4]
[1*]"Vasile Goldis" Western University, Department of Paediatrics, Arad
[2,3] County Emergency Hospital, Department of Paediatrics, Arad
[4]"Luis Turcanu" Children Hospital, Timisoara

ABSTRACT. Achondroplasia is a rare autosomal dominant genetic disease and the most common form of short-limbed dwarfism. The clinical achondroplastic features are: short stature and limbs, enlarged head with prominent forehead, lordosis and a trident aspect of the hand. The intelligence is usually normal. Because of the complexity of the complications (apnea, motor retardation, hydrocephaly, respiratory disorders, spinal stenosis, spinal cord compression) a child with achondroplasia has to be periodically monitored by a multidisciplinary team, especially in the first years of life. Moreover, the role of the psychological counseling is essential for a child with achondroplasia and his family. Also, a better communication network among different specialists and the support associations is needed, in order to create a professional guidance and a normal environment for those who are living with achondroplasia.

Keywords: achondroplasia, short stature, limb-lengthening, spinal stenosis, multidisciplinary team

INTRODUCTION

Achondroplasia is a rare autosomal dominant genetic disease and the most common form of short-limbed dwarfism. It occurs in 1 in 10,000 to 30,000 newborns and 80% of cases seem to be the result of a spontaneous mutation (Horton WA et al., 2007). The key in the pathogenesis is a mutation in fibroblast–growth–factor–receptor 3 (FGFR3) in a gene located on the chromosome 4, which causes the inability to form bone from the cartilage, by an abnormal endochondral ossification (Tanaka H et al., 2010).

Because of the complexity of the complications that can occur, especially in the first years of life (apnea, obesity, motor retardation, respiratory disorders, hydrocephaly, spinal stenosis, spinal cord compression) a child with achondroplasia has to be periodically monitored by a multidisciplinary team that should include a paediatrician, paediatric neurologist, paediatric neurosurgeon, orthopedic surgeon, endocrinologist, genetician etc. It is essential that a psychologist should also be included, as the counseling could help the child and his family to live a normal life. Moreover, more medical involvement is needed for helping the support associations in their tasks to provide psycho-social strengthening, for sharing the medical knowledge and for creating a better understanding of life with achondroplasia.

DIAGNOSIS

Achondroplasia is diagnosed based on the clinical appearance and on the molecular confirmation of the specific mutation (FGFR3G1138A).

The clinical achondroplastic features are: short stature (average height is 131 cm for a male adult and 124 cm for a female adult), short limbs with particularly short upper arms and thighs (rhizomelic dwarfism), enlarged head (macrocephaly) with a prominent forehead and flat nasal bridge, lumbar lordosis or kyphosis. Another characteristic feature is the trident appearance of the hand (a wide space between the middle and the ring finger). The intelligence is usually in the normal range.

The prenatal diagnosis can also be suspected by ultrasound when excessive amniotic fluid, small limbs or an enlarged head can be detected. A karyotype analysis is performed in these situations. If the karyotype analysis does not explain the abnormal sonographic findings, a mutation analysis is performed. (Ros –Pérez P et al., 2012).

THE ROLE OF A MULTIDISCIPLINARY TEAM

The diagnosis of a child with achondroplasia has a major impact on his family and, therefore, the role of a multidisciplinary team is very important. The psychological support of the family must be a priority as well as the medical monitoring of the child, as achondroplasia can have complex complications, where prompt intervention is required to save the life (neurosurgical decompression of the spinal cord).

THE ROLE OF THE SUPPORT ASSOCIATIONS

As mentioned above, the support associations play an important role in providing psycho-social guidance, job orientation and society integration programs for these children and their families. Unfortunately, height remains a criteria of judgement in our society. To have a job or to be an equal member in the society are fundamental needs and no one should be isolated because of some old fashioned prejudices.

TREATMENT

Although achondroplasia has no specific treatment and the main hope remains the genetic therapy (antibody blockade of receptor activation, alteration of pathways that modulate the FGFR3 downstream signaling - Lorget F. et al., 2012), there are medical

**Correspondence:* *Simona Dumitra, "Vasile Goldis" Western University, Department of Paediatrics, e-mail: dumitrasimona@yahoo.com*
Article received: April 2013; published: September 2013

and surgical options to correct some of the complications. The children with achondroplasia often face psychological trauma because of their appearance and for being "different" (short stature with disproportionally short limbs), so they can be offered some medical or surgical solutions. The medical solution for increasing the height is the growing hormone therapy. This therapy is, usually, recommended in the first years of life with the most accelerated rate of growth observed in the first year after the onset of the therapy (Seyno Y, 2009). The surgical solution, though controversial, is the limb-lengthening surgery that lengthens the legs and the arms - it can increase the height by as much as 30 cm. For some, the question of whether or not to perform limb lengthening, is a question of letting these people be more independent. Complications due to surgery can occur and sometimes repeated surgery is needed. It is hard for parents to decide, even with professional help, thus involvement of the child in decision-making at the start of the treatment is expected and recommended from the age of 12 (Schiedel F et al., 2012).

Another complication that is both common and potentially lethal in children with achondroplasia (and where an early diagnosis and treatment can make the difference between life and death) is the cervico-medullary compression. It is associated, if not promptly treated by neurosurgical decompression, with severe morbidity and even sudden death, especially in infants 4 years of age or younger (Ozcetin et al., 2012).

Children with achondroplasia can also develop sleep disorder problems (apnea, hypopnea). Therefore, due to their high prevalence, systematic sleep studies are recommended as part of the routine annual evaluation. If the polygraphic sleep studies are pathological, upper airway surgery and noninvasive positive pressure ventilation (NPPV) are effective therapeutical solutions (Julliand S et al., 2012)

Though achondroplasia may have severe and sometimes lethal complications, the majority of children and adults can live a normal life. This is an important fact to be aware of as a doctor when treating a child with achondroplasia as their characteristics should be known and respected.

CASE REPORT

In the following report, we are going to present the story of a 4 year old boy who, although diagnosed with achondroplasia and non-progressive foramen magnum stenosis, lives a normal life due to the continuous efforts of his family.

The boy was born on the due day and had normal anthropometric parameters: birth weight =3400g (P50-75%), birth length=48cm (P10%) and head circumference=33cm (P10). During pregnancy, the ultrasound findings were normal and there were no alarming events. He is the second son of a non-consanguineous couple. There was no history of relatives with short stature and his older brother is healthy.

At the age of 6 months old, a larger head circumference (P>90%) and a non-concordance between the body and the limbs length was noticed. Thus, a head ultrasound was performed and hydrocephaly was suspected. Consequently, a MRI scan was performed, which excluded the hydrocephaly and showed an atypical foramen magnum for the child's age. At this point, based on the clinical and radiological findings, the diagnosis of achondroplasia was given. As the neurological examination revealed a normal tonus and development, there was no neurosurgical indication for foramen magnum decompression at the time. The boy undertook two more MRI scans, 6 months apart, which showed a non-progressive foramen magnum.

The genetic tests were done at the age of 2 years old and confirmed the diagnosis of achondroplasia caused by a spontaneous mutation.

DISCUSSION

As the diagnosis was found, questions were asked about treatment and about facing the consequences of achondroplasia. As the repeated MRI scan showed a non-progressive foramen magnum, no further neurosurgical follow up and investigations were needed, but if new neurological symptoms should appear, the situation should be reviewed.

The boy also developed genum varum and he has yearly orthopaedic checkups as in some severe cases the genum varum needs surgical correction.

Faced with such a severe diagnosis, the family found support from some professional doctors, but these represented only a small minority, insufficient to meet the family's expectations and needs in their difficult and challenging journey.

They also found help from The Association of Small People and The Association Integra in Romania and became very active members. Moreover, the support associations provided some financial help, as the boy needed and still needs medical follow ups that sometimes involve visits to various clinics in Romania and abroad. Also, he should have (daily) physiotherapy and swimming sessions, but the costs are prohibitive, even for a middle class (Romanian) family.

Through the help of the support associations, the parents went to Germany where they met the BKMF group (German association of short-statured people and their families). Here, they found more professional guidance and especially more understanding and empathy than in their own country and, as hard as it is to believe, than in their own family. They were very much impressed by everyone's involvement (doctors, nurses, psychologists, surgeons) in providing a normal environment and in finding the best medical and social options for the patients.

CONCLUSIONS

By sharing the difficult journey of this wonderful family, the authors would like to raise awareness of this disease. Also, they would like to challenge the medical community and the relevant authorities to improve the support offered to the achondroplasia patients and their families. Better guidance, better communication is acutely needed, ideally from a multidisciplinary network of medical specialists and support associations.

REFERENCES

Julliand S, Boulé M, Baujat G, Ramirez A, Couloiner V, Beydon N, Zerah M, di Rocco F, Lemerrer M, Cormier-Daire V, Fauroux B, Lung function, diagnosis, and treatment of sleep-disordered breathing in children with achondroplasia, Am J Med A. 2012 Aug;158A(8):1987-93.

Horton WA, Hall JG, Hecht JT, Achondroplasia, Lancelet. 2007, Jul 14;370(9582):162-72.

Kim SJ, Balce GC, Agashe MV, Song SH, Song HR, Is bilateral lower limb lengthening appropriate for achondroplasia?: midterm analysis of the complications and quality of life, Clin Orthop Relat Res. 2012 Feb; 470(2):616-21.

Lorget F, Kaci N, Peng J, Benoist-Lasselin C, Mugnierv E, Oppeneer T, Wendt DJ, Bell SM, Bullens, Bunting S, Tsuruda LS, O'Neill CA, Di Rocco F, Munnich A, Legeai-Mallet L, Evaluation of the therapeutic potential of a CNP analog in a Fgfr3 mouse model recapitulating achondroplasia, Am J Hum Genet. 2012 Dec7;91(6):1108-14. doi: 10.1016/j.ajhg.2012.10.014. Epub 2012 Nov 29.

Ozcetin M, Arslan MT, Karapinar B, An achondroplasic case with foramen magnum stenosis, hydrocephaly, cortical atrophy, respiratory failure and sympathetic dysfunction, Iran J Pediatr 2012 Mar;22(1):121-4.

Ros –Pérez P, Regidor FJ, Colino E, Martinez-Pavo C, Barosso E, Heath KE, Achondroplasia with 47, XXY karyotype: a case report of the neonatal diagnosis of an extremely unusual association, BMC Pediatr. 2012 Jun 2012 29;12:88.

Schiedel F, Rodl R, Lower limb lengthening in patients with disproportionate short stature with achondroplasia: a systematic review of the last 20 years, Disabil Rehabil. 2012;34(12):982-7.

Seino Y, Updated treatment of achondroplasia, Clin Calcium. 2009 Mar;19(3):432-6. doi: CliCa0903432436.

Tanaka H, Cytokines in bone diseases. FGF receptor signaling and achondroplasia/hypochondroplasia, Clin Calcium. 2010 Oct;20(10):1490-6.