Witness an online magazine

Achondroplasia

Casting Away The Stigma

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Achondroplasia is the most commonly recognized form of dwarfism.

It doesn't choose gender, race, or ethnicity. It is a form of skeletal dysplasia that affects approximately one in every 40,000 children in the world; one in every 10,000-15,000 children in the United States, and in Spain, where this story was photographed, one in every 3,000.

Achondroplasia is usually detected in the utero or at birth. It is caused by a genetic change or a mutation in a genetic instruction called FGFR3, the fibroblast growth factor receptor, which results in an abnormality of cartilage and bone formation. It can happen to anyone in the general population.

A dwarf is described as a person of short stature who stands at 4 foot - 10 inches (147cm) or under.

There are many challenges associated with life as a "little person," from possible early-life complications to social stigmatization.

Even the connotation of the term, "dwarf," can vary dramatically from person to person.

Noah in the doctor's office.

"Dwarfism is a term still applicable to patients with severe short stature and still accepted by most of the medical community and even by family support groups. Some people, however, find the term rather pejorative and prefer either 'bone dysplasia,' 'skeletal dysplasia,' or 'little person,'" says Dr. Carlos Bacino, M.D., a clinical geneticist and cytogeneticist. Bacino is the chief of Genetics Service at Texas Children's Hospital and professor of Molecular and Human Genetics at Baylor College of Medicine in Houston, Texas.

Dr. Ingrid A. Holm, M.D., assistant professor of pediatrics at Children's Hospital Boston, whose concentration is in clinical research and genetics, is among those who prefer the term "skeletal dysplasia."

"I actually tend not to use the word dwarfism. I don't know if other people do. I don't really like that word. It gives some types of funny connotations," says Holm. "There are many, many types of skeletal dysplasias – bone defects – and achondroplasia is probably the most common of these genetic disorders that primarily affect bone growth and development."

In the schoolyard, Ot (lower right on toy car), plays with the other kids. Teachers are careful and show some deference in class, but outside he is just like any other kid.

Ot's classmates are aware of his situation and always give him extra help.

Fighting against the stigma and discrimination associated with being a person of short stature is still the greatest challenge that dwarves, achondroplasiac or not, have to face on a daily basis.

"Little People of America" is a nonprofit organization dedicated to improving the quality of life for people with dwarfism while also celebrating with great pride their contribution to social diversity, says Leah Smith, LPA vice-president of Public Relations.

The organization has over 6,000 members, both nationally and internationally, and several sister organizations in other countries.

"Little people were traditionally portrayed in circuses and sideshows; however, we are now trying to move away from this stigma to becoming professionals," says Smith. "We have members who are doctors, lawyers, professors, accountants, architects, etc. However, this stigma is still one we face on a daily basis. We have had Harvard graduates who graduated at the top of their class report having faced employment discrimination."

"This stigma permeates not just employment, but many aspects of our lives and often produces a great deal of ridicule. This

On Sundays, Ot usually goes for a walk near the beach with his family. Sometimes people can be tactless. Their stares are sometimes annoying and that is something that he and his family must accept.

Ot plays with his 8-month-old sister in his room with his parents.

is a very real problem and one we try to overcome on a daily basis on an individual and organizational basis."

According to medical research, the average height for an adult female dwarf is around 4'1" (125 cm), ranging from about 3'8" to 4'7" (115 to 140 cm), and approximately 4'3" (130 cm) for the average adult male, ranging from 3'9" to 4'8" (120 to 145 cm). There are over 400 types of dwarfism, with some being extremely rare and others more frequent.

People with achondroplasia have several distinctive physical characteristics that can cause medical complications. But with consistent medical follow-ups with a geneticist and orthopedic specialist over time, they can have a normal life.

Individuals with achondroplasia have "a large head with prominent forehead; underdevelopment of the mid-part of the face; shortening of the upper and lower extremities, with the shortening more noticeable in the parts of the limbs close to the shoulder and hip; increased curvatures of the spine; short hands and feet; and moderate laxity (looseness) of the large joints," says Bacino.

It is this shortening in certain parts of the body and not others that gives achondroplasiac dwarves some appearance of disproportion.

Most of the medical issues affecting an achondroplasiac child occur in the first few years of life. After that, it is important to continue to monitor growth while looking out for any other complications.

"The treatments for achondroplasia are mainly supportive and aimed to help with possible bone complications in the lower part of the head, compression of the spine, and bowing of the bones," says Bacino. "In the first few months of life up until 1-2 years of age, we carefully monitor head growth, since the base of the skull may grow very slowly and that can cause problems with chord compression. Later on, in adult life, we monitor the lower part of the spine, also due to possibilities of narrowing."

Helping parents understand what to expect and how to react early on is key.

Children with achondroplasia can have problems with their neck, so the parents need to be very careful to support their heads until they are strong enough to do it themselves.. They can also have sleep issues and apnea because of the way the airway and the air entrance to their lungs is shaped, explains Holm.

"So, if they have respiratory chronic issues, I'll send them to have a sleep study to see if they need help to sleep at night or need to have their tonsils removed," says Holm.

People with achondroplasia have a normal IQ, but as kids, sometimes their physical problems can lead to difficulties in learning.

This happens because they are often more worried about their stature than about their studies. It can also be due to otitis (inflammation or infection of the ear), low class attendance, or a hindrance to their motor development caused by the bigger proportion of the cranium in relation to the body. All of this can be solved with early stimulation, so it's important for a family to have a good communication with the child's doctors and school.

"[Kids with achondroplasia] don't have developmental issues, so that helps," says Holm. "They don't have cognitive issues, they don't have the retardation, or delay. They have some birth motor delays, but cognitively they do fine. And as they get older, they have to deal with their size, but you have a lot of hope for them."

Ot feeds his fish. He climbs chairs and furniture all over the house to reach his objectives.

After recess, the children make a line. Here the difference in height is clear.

At first look, Ot is like any other energetic 4-year-old boy watering his garden with a hose or playing outside with his friends. It's not until he lines up next to other boys his age that the difference in his height is made obvious.

Ot is an achondroplasiac dwarf, but he doesn't look like a typical achondroplasic kid. His facial features are quite slim, but his limbs and his stature are short.

He is aware that he is smaller than others, but that doesn't usually bother him. His lack of stature is compensated by other skills, such as his persuasive nature with both adults and friends. There are

times when he tries to get extra attention by asking for more help than he really needs, but that's typical of any child.

With the right medical care and some adaptation, the life expectancy of a dwarf is completely normal. Adjustments that need to be made vary and include basic necessities that most people take for granted, such as bedding, seating, and bathroom and school accessibility.

At home and at school, Ot has adapted spaces for him, such as the toilet, the blackboard and the chair at his class. He has a little step to rest his feet. The doors also have to be adapted because he cannot reach the handles. Whenever they leave the classroom, the teacher holds Ot's hand to help him avoid any unnecessary falls or blows. The rest of his daily life is no different from that of any other kid.

"Most people with short stature adapt appropriately, but there is definitely a struggle for them to fit in the everyday living," says Bacino. "I have not seen significant problems with self-esteem over the years, but it is definitely an issue that most adolescents deal with when growing up with such disability. It's common to hear parents telling me that their children are being picked on or being made fun of."

A feeling of inclusion in the group helps children with achondroplasia feel accepted in their environment.

Ot is lucky. Though his classmates know he is different, they help him and look after him.

In his house, Ot is king. He loves helping his father fix his motorcycles in the garage. His dexterity makes up for some other lacks – things he can't do due to his stature or age. Following his father's example and feeling useful help him build character and confidence.

Ot has a sister who is average in stature. Although he is three years older, she is already taller than him. But he doesn't care and plays with her as he always has.

His family is very supportive and tries to help him live a normal life. Their greatest challenge is to offer as much normality and strength as possible, while also setting an example of behavior in difficult situations.

In his little adapted bathroom the difficulties of Ot's daily routine are clear. For adults or children, their stature hampers their everyday life.

Noah training with his team, C.F. Olímplic la Garriga. He trains twice a week.

It's important for a child of short stature not to feel victimized. Finding a healthy balance that allows the child to feel safe but not overprotected can be a challenge. Achondroplasia support groups and physicians work with the individuals and their families to offer this type of support.

Treatment for achondroplasia doesn't change significantly from country to country. As with anything else, nations with a better health care system can provide access to more surgical modalities.

Though most of the treatment for achondroplasia is supportive, there is a surgical procedure that can help little people add a few extra inches to their height. It is a limb lengthening procedure that follows a surgical technique popularly known as the "Ilizarov technique."

Bacino says, "The process is long and requires a surgical break that is done in the femur and humerus. It's done to one set of limbs at a time, and after the breaks, some hardware is placed that needs to be changed frequently to stretch the bone and make room for more. Each set of limbs takes at least one year and when done in the lower limbs requires immobilization so the patients need to be on a wheelchair for an entire year. The entire process takes 2 years, is extremely costly, and not favored by many. It is done after growth is completed (after puberty) and requires full cooperation and adjustment to the process. A minority of people go that route."

Although the procedure is long and painful, some people do choose to go that route. The importance of those few extra inches or centimeters can be immeasurable. The extra height provides people of short stature with more independence and can even make the difference between reaching a door handle, an ATM machine, or an elevator button or not.

Every year, more than a hundred kids between the ages of 10 and 14 face tough operations with adult-like strength, putting all of their hopes in the possibility of reducing their difference.

Noah, an 11-year-old teenage boy from Spain, has undergone the procedure to lengthen his bones. Until just a few days before his surgery, Noah was running after a soccer ball at full-speed.

Soccer is his favorite sport. He trained with his team, the C. F. Olímpic, but couldn't actually compete due to his height. That has never kept him from feeling like a part of the team. His teammates adore him because of his tenacity, support, and dedication.

Noah with his teammates.

A person with achondroplasia can't really practice sports at a competitive level or practice sports based on physical strength, such as gymnastics and acrobatics. But there are many other sports that can help them strengthen their muscles, like swimming.

When playing soccer, Noah needs to be careful and to avoid hitting the ball with his head, due to spinal stenosis. He is an achondroplasic, but that is only one of his characteristics. He has many more.

Noah is very sociable, easy-going, and well-loved by those in his environment. Wherever he goes, he is known and valued for who he is and for his personality – not for his height.

Before the surgery, Noah was 3'7" (110 cm) tall. The purpose of the procedure is to give him an extra 6 inches (15 cm) of height to help improve his quality of life as an adult and prevent possible malformations.

As he says, with his excellent sense of humor, "Being taller will make me more successful with girls."

Noah is helped out of bed.

The X-ray shows the growth and the correction of the legs curvature. The development of the bones must be carefully monitored for the entire process.

The growth of his shinbone is checked at the hospital's by the medical staff. Every day the bars must be turned until they reach 15 cm.

Like many other kids, Noah has been preparing to face this procdure for a long time. From his birth, the follow-ups at the hospital have been constant and have played a key role in his life.

At the Institut Dexeus in Barcelona, Spain, where Noah's procedure was done, they work to make the process as comfortable and easy as possible. At the Growth Unit, they provide support to the children and their families by explaining all of the problems and difficulties of the treatment.

In the case of children, the doctors insist that they continue studying. The parents have to show unconditional support while taking the doctors' advice.

The children are kept in touch with other people with skeletal dysplasias who have undergone the procedure, which helps them understand the treatment and not be afraid of it. It's a step-by-step process and making sure that the individual is psychologically prepared for it is crucial.

Noah has received news from his doctor that he must stay two more days in the hospital. He sheds some tears while his mother tries to comfort him.

After all the medical studies on the legs, their inclination, and their way of walking are done, comes the big moment – the surgical procedure.

Since his surgery, Noah's independence has been greatly affected. His movements are much more limited and he always needs to be accompanied by someone and his wheelchair.

During the first months, the pain can be especially intense. When he needs to be transferred from place to place, two people are needed to move him. Everybody needs to be patient because Noah's first movements are extremely difficult because of the pain.

On the first days of the surgery, Noah's pain was so great that he wanted to have the bars removed. The bars need to be unscrewed

The bars and skin must stay clean in order to avoid infection.

one millimeter every day. One millimeter may seem like little, but at the end of the entire process, the six inches (15 cm) desired will be reached.

Balance comes little by little and pain becomes a habit, especially when dressing the wounds. With the help of a walker, Noah can start walking just a few months after the operation, but even a few steps can seem to take forever. In this phase, patients are ashamed of walking down the street and of being seen doing the exercises they need. Psychological rejection is common, but Noah tries his best to behave naturally.

During the last months after the surgery, the anxiety to remove the bars and walk again can be huge. A balanced and supportive family environment is imporant to help with adaptation and reinforce self-esteeem. Noah's family tries to keep him occupied so that he won't get bored or discouraged. His friends always visit him too.

Noah enjoys watching his legs grow. His calves have stretched quite a bit, so he will need to strengthen his muscles and quadriceps

Exercise is very important to improve the strength of the legs. Six months after the operation, Noah needs to stand up. Pain appears immediately, but walking helps the extension process.

Every week Noah visits the physiotherapist to strengthen his muscles. He needs to learn to walk with his new height, because his legs have grown 15 centime-

so that they will hold his weight.

After the bars have been removed, Noah needs to rest and keep his legs in plaster for a few months. Then, it's back to walking and strengthening his muscles with the help of a physical therapist on a weekly basis.

Noah smiles whenever he remembers playing soccer with his friends. That's what keeps him going throughout the long and painful process – a positive outlook and the thought of being out on the field again.

With the advancement of technology and medical research,

other types of surgical and supportive treatments will become available for those who want it.

"Right now, there are some exciting treatments based on research that has been known for a number of years," says Bacino. "This treatment will soon be part of trials through a commercial company called Biomarin. There is a drug called C-type Natriuretic Peptide (CNP), that exerts a powerful effect over the long bones and there have been animal studies that showed that CNP can lengthen the long bones in animal models with achondroplasia."

The drug is called BMN-111 and Baylor College of Medicine will be one of the sites for the studies. According to Bacino, they will start recruting patients for Phase I and Phase II of the trial some time in 2012. The details are still being worked out.

Despite their differences, "little people" are like anyone else – people with hopes, fears, and dreams trying to live a life of acceptance and meaningfulness. Oftentimes, they require even more perseverance than those without a disability as they fight for the right to be seen as equals.

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ADDITIONAL RESOURCES

Little People of America
www.lpaonline.org

Baylor College of Medicine
http://www.bcm.edu/genetics/index.cfm?pmid=10580

Children's Hospital Boston
http://www.childrenshospital.org/cfapps/research/data_admin/Site283/mainpageS283P0.html

BioMarin
http://www.bmrn.com/pipeline/cnp-for-achondroplasia.php

This project is a production of

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