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SHORT REPORT

Achondroplasia Day 2012 in Johannesburg

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The Division of Human Genetics (National Health Laboratory Service and University of the Witwatersrand) recently hosted the second annual Achondroplasia Day in Johannesburg. Participation in the meeting increased from 4 families in 2011 to 17 families in 2012. The main aim of the meeting was to bring individuals with achondroplasia and their families together and to facilitate improved interaction between families and the healthcare professionals involved in their care. This short report briefly describes the events of the day, and the immense benefit such activities have for families with rare genetic conditions. Additionally, we provide an overview of the basic genetics of achondroplasia and specific health needs of affected individuals. It is hoped that other centres around South Africa will be inspired to organise similar events in their respective areas to benefit their patients.

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Achondroplasia Day

The Division of Human Genetics (National Health Laboratory Service (NHLS) and University of the Witwatersrand) recently hosted the second annual Achondroplasia Day, held at the Rahima Moosa Hospital in Johannesburg. Participation in the meeting increased from 4 families in 2011 to 17 families in 2012. The main purpose of the meeting was to bring affected individuals and their families together and to facilitate increased interaction between families and the healthcare professionals involved in their care. Holistic care for patients involves not only treating their biological or physical symptoms, but recognising their psychological and social needs as well. We therefore hoped to provide a platform for families from all walks of life to share their experiences and provide support for one another. In addition to the genetics team, the meeting was attended by physiotherapists, occupational therapists, nurses and medical students. The programme included informal talks by adults with achondroplasia, talks on the role of physiotherapy and occupational therapy in achondroplasia, and a medically orientated presentation on the basic genetics and health concerns faced by people with achondroplasia. This was especially helpful for parents who had never received genetic counselling, and was an excellent opportunity to reiterate the most important issues.

Basic genetics and clinical findings

Achondroplasia is the commonest form of short-limbed dwarfism, affecting 1 in 26 000 - 28 000 newborns worldwide, with no predilection for gender or ethnicity.[1] It is an autosomal dominant condition, caused by mutations in the *FGFR3* gene, located on chromosome 4. Approximately 80% of children with achondroplasia are born to average-statured parents, due to new (*de novo*) mutations. In such cases, the recurrence risk for future pregnancies is low. However, if one parent has achondroplasia, there is a 50% risk in each pregnancy of having a child with achondroplasia. Should both parents be affected, the risks to their offspring are as follows: 25% of having average stature, 50% of having achondroplasia, and 25% of having homozygous achondroplasia (a lethal condition). Achondroplasia may be suspected on prenatal ultrasound, and is evident at birth. The diagnosis is reliably made on clinical and radiological grounds (Table 1), but genetic testing may be needed in certain cases[1] (testing is available through the NHLS).

Table 1. Clinical and radiographic features of achondroplasia

Clinical features	Radiographic features
Disproportionate short stature	*Long bones*: Short, robust tubular bones; generalised metaphyseal changes (may be mild)
Large head with frontal bossing	*Spine*: narrowing of the interpedicular distance in lumbar spine
Mid-face hypoplasia with depressed nasal bridge	*Pelvis*: rounded ilia and horizontal acetabulae; narrow sacrosciatic notches
Rhizomelic shortening of the arms and legs	*Femurs*: proximal radiolucency of femoral heads
Brachydactyly, often with trident configuration of the hands	
Bowed legs	
Thoraco-lumbar kyphosis in infancy	
Exaggerated lumbar lordosis when ambulatory	
Normal/average intelligence	

Specific health issues faced by individuals with achondroplasia

The American Academy of Pediatrics (AAP) has a specific health supervision guideline for people with achondroplasia,[2] which includes achondroplasia-specific growth charts. Suggested regular surveillance includes monitoring of:

- growth, including head circumference (for hydrocephalus during infancy)
- signs of upper airway obstruction (due to mid-face hypoplasia)
- abnormal neurological signs (spinal/cervico-medullary stenosis)
- signs of frequent ear infections (which may compromise hearing).

Resource constraints may make it impossible to adhere strictly to all the AAP recommendations. However, even seemingly trivial advice given to parents can significantly influence the quality of life of people with achondroplasia. For example, we consistently advise against certain practices such as back-carrying of infants with achondroplasia. Back-carrying is common in South Africa, but it increases the risk of gibbus formation and subsequent neurological impairment in these children. Care for 'head, neck and back' needs to be reiterated at every health visit. Equally important aspects such as the practical difficulties associated with short stature, and the psychological impact of achondroplasia, should be addressed. This is where meetings such as this one are important, as well as the establishment of parent support groups.

Feedback from participants

For the majority of parents, this was the first opportunity to meet with other families. The feedback from all participants was overwhelmingly positive. The adults with achondroplasia, some of whom also had children with the condition, were able to offer invaluable advice on practical day-to-day issues, such as where to get suitably sized clothing and shoes. One mother realised for the first time that her daughter would be able to have children of her own. An 8-year-old boy with achondroplasia asked his father whether there really were 'mummies and daddies' that were as small as him, as he marvelled at the articulate speaker with achondroplasia. The meeting served to confirm what we as doctors have been telling parents all along: people with achondroplasia can lead fulfilling lives and contribute positively to society.

We hope that this successful initiative will inspire other centres around the country to organise similar events to benefit patients with achondroplasia and their families.

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References

1. Pauli RM. Achondroplasia. 1998 Oct 12 [Updated 2012 Feb 16]. In: Pagon RA, Bird TD, Dolan CR, et al., eds. GeneReviews™ [Internet]. Seattle (WA): University of Washington, Seattle; 1993. http://www.ncbi.nlm.nih.gov/books/NBK1152 (accessed 1 March 2012). [PMID: 20301331]
2. Trotter TL, Hall JG. American Academy of Pediatrics Committee on Genetics; Health supervision for children with achondroplasia. Pediatrics 2005;116:771-781. http://dx.doi.org/10.1542/peds.2005-1440 (accessed 1 March 2012). [PMID: 16140722]