Requests for additional copies can be sent to:

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**FORWARD**

When first told that their baby has achondroplasia, many parents feel almost overwhelmed with questions and worry. In addition to medical concerns, like other parents, you may wonder: “How will my child learn to do the things other children do?” “When will my child learn to sit, to crawl, to walk, to dress and undress?” “Is what we are seeing normal or a sign that something is wrong?” This booklet was written to allay fears and answer questions about development in infants and children with achondroplasia. It focuses on the characteristic developmental patterns and movement strategies we have observed in these children. While your child’s development may, in fact, be different from the developmental patterns seen in average statured children, ‘different’ does not mean ‘defective’. Realizing that many of the developmental differences you may observe are adaptations to physical differences should alleviate some of the anxieties that parents and caregivers may have.

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Much of the information contained in this booklet is based on nearly a thousand clinical assessments of children with achondroplasia at the Midwest Regional Bone Dysplasia Clinic of the Clinical Genetics Center, University of Wisconsin-Madison. In addition, some information about movement strategies derives from a specific parent questionnaire. More details can be found in Fowler ES, Glinski LP, Reiser CA, Horton VK and Pauli RM: Biophysical Bases for Delayed and Aberrant Motor Development in Young Children with Achondroplasia, *Journal of Developmental and Behavioral Pediatrics* Volume 18, June 1997.

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**INTRODUCTION**

Achondroplasia is one of a fairly large group of inborn characteristics that result in abnormalities of bone growth. This group, as a whole, is referred to as *skeletal dysplasias* or *bone dysplasias* or *osteochondrodysplasias*. Achondroplasia is the most common of these bone dysplasias, occurring about once in every 25,000 newborns. The basic, molecular cause of achondroplasia recently has been discovered: achondroplasia arises because of a change in a specific gene, the Fibroblast Growth Factor Receptor Type 3 (FGFR3). This gene codes for a protein that is crucial for certain cells to recognize growth stimuli and to translate those signals into one that the inside of the cells can recognize. The specific change of one copy of the FGFR3 gene found in nearly all individuals with achondroplasia either directly or indirectly causes all of the effects seen in affected persons.

Children with achondroplasia are usually cognitively normal — that is, they usually have normal brain function, do well in school and can pursue virtually any career. However, concerns often arise because a child with this diagnosis may show considerable delays in development early in life. In fact, for many motor skills substantial lags can be expected. In addition, most children show unusual (‘aberrant’) patterns of motor skill attainment.
Physical Features That Can Affect Motor Development

All children with achondroplasia have shortening of the long bones of the arms and legs, particularly of the thighs and of the upper arms:

- Thigh shortening
- Short upper arms

In addition, there is usually decreased muscle tone (that improves as a child gets older), a large, heavy head, and limitation of elbow extension:

- Limited elbow extension

Loose joints can also affect motor development. These two children show the marked hip and knee hypermobility that can result in unusual positions and postures:

Children with achondroplasia also have short fingers with increased separation between the middle and ring fingers (often referred to as the ‘trident configuration’ of the hand).

Although most children with achondroplasia display most or all of these features, the severity of each characteristic differs from child to child.