RESPIRATORY STUDIES IN ACHONDROPLASIA DURING CHILDHOOD

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Twenty years ago, infants with achondroplasia were believed to have a high death rate. The reason most often given for this was difficulty with breathing because of an unusually small chest cage which did not allow for enough expansion of the lungs. As diagnostic ability began to improve and other conditions could be separated from classic achondroplasia, this attitude began to change. More recently, achondroplastic infants and children were believed to have few health problems in comparison to those affected by most other forms of short stature. It was believed that most of these babies were generally healthy except for the uncommon occurrence of increased fluid pressure on the brain due to hydrocephalus (impaired drainage of cerebrospinal fluid "water on the brain") or rare instances of paralysis from compression of the upper spinal cord because of constriction by the bones of the neck and the base of the skull.

In 1983, doctors at Johns Hopkins reported that a certain group of infants and children with achondroplasia had chronic breathing problems, due to three types of factors related to the condition: (1) small chest cage, (2) problems with breathing during sleep due to constricted nasal passages and (3) abnormal breathing patterns due to chronic compression of the upper spinal cord. The first two appeared more commonly and were also more easily diagnosed. About the same time, it was noted by a number of medical advisors to LPA that there was a disproportionately large number of sudden unexplained deaths among achondroplastic infants. The following year, they reported the results of their study of the problem, concluding that, despite a lack of clues prior to the deaths of these babies, spinal cord compression was potentially an important contributor to their deaths. It was suggested by the Medical Advisory Board that research studies to look into this problem be undertaken. Based on our prior experience at Johns Hopkins with respiratory problems in achondroplasts of different ages, we intensified our studies in younger children, continuing our focus on respiratory problems. The newer ideas generated through the Medical Advisory Board led us to expand our study to include evaluation for possible compression of the spinal cord in all patients as well, regardless of whether they had symptoms. However, there was a need to establish the normal range of expectation and to define what would be considered outside the range and in need of treatment.

We began by trying to evaluate how the size of the chest varies with age, since it was known that some babies with very small chests had symptoms, such as rapid breathing and recurrent pneumonia. We also sought to find how chest size related to symptoms. Therefore, beginning in 1983, we began to measure as many achondroplastic children under age 7 as possible. Within three years, with the cooperation of LPA, over 100 different children were measured. Over the next 7 years, a number of children have had repeated measurements to assess their chest growth as well. In each case, parents were asked about symptoms that might indicate respiratory problems. Based on the survey, we learned that small chests are very common in those younger than 2 and that, when specific questions are asked, nearly half of the babies have symptoms of a breathing disorder, such as chest infections or rapid breathing. Very few, however, were seriously ill and most babies appeared to tolerate the small chest well. Moreover, the chest sizes of children over two were within the range for average-sized babies, although they were always less than the average. Even babies who had earlier symptoms we thought were due to a small chest grew out of them by age 2 or 3. Since this was a relatively small survey, it is possible that we measured a group which is not representative of all babies with achondroplasia; therefore, we may have over- or under-estimated the risk that a specific baby with a small chest would have symptoms.

We also looked more closely at a group of children who either had specific symptoms or were entered into the study by interested parents. These 27 children were evaluated medically to determine their risk to have the three problems identified earlier as causes of respiratory symptoms. The first thing we learned was that some children who had been thought to be "well" actually were having significant symptoms when specific questions were asked. Parents had been accustomed to being reassured that rapid breathing or having sleep apneas (stopping breathing for varying periods) was normal for their child. Evaluation of these children showed that same of those who had these symptoms had strain on their hearts from chronically or intermittently law oxygen levels. We also noted that there had been reassurance regarding children who had significant delay of their motor milestones, such as standing and walking, beyond the expectation for achondroplasia. Same of these children were neurologically abnormal and really had weakness. We also learned that CT scan X-rays of the area where compression occurs were sometimes abnormal in children with no neurologic symptoms and respiratory studies were sometimes abnormal in children who had no respiratory symptoms. These and other findings convinced us of the difficulty in assessing any individual child without complete evaluations. In all, we found that 23 of 27 children had medical problems causing breathing abnormalities—10 with spinal cord compression and 13 without spinal compression. On the other hand, two children whose parents believed they had breathing problems had normal tests. All ten children with compression had surgery. We learned how difficult the surgery was and the kinds of complications, such as infections, which could occur. Although two children did not do well, the others who had surgery for their compression have had good results and are doing well several years later.

Dr. Cheryl Reid

Since we published the results of our study in 1987, children with achondro-

plasia who have respiratory problems continue to be identified. There have been some changes in how they are managed. At Cooper Hospital/University Medical Center, we have formed a multidisciplinary team for children with skeletal dysplasias, including achondroplasia. Each child in the program is seen by several specialists in a single visit and a plan for evaluation and management is developed together by the team. A similar system has emerged at other medical centers with special interest in achondroplasia. Many of these centers now have sleep laboratories and respiratory specialists devoted to the care of children, so that specialized lung studies that were not possible earlier can now be done. We have begun to use magnetic resonance scans (MR) in addition to CT scans to evaluate the base of skull for compression and have learned that both studies seem necessary to fully evaluate each child who needs evaluation. The earlier work has shown that a good history obtained from a family still allows us to identify children who most need evaluation as well as those whose families can be reassured. Many of the questions about respiratory problems in achondroplasia are likely to be answered with the continuing study that is being undertaken. ◆

Editor's Note: Dr. Cheryl S. Reid is a member of LPA and of the Medical Advisory Board.

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**Joan was accidently left of the MAB list in the last issue, my apologies. Also, please note this is a new mailing address for Joan and the Alliance.*