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Case Report – Achondroplasia

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Abstract

Achondroplasia is the most common form of skeletal dysplasia resulting in disproportionate short stature and affects over 250,000 people worid wide . we here present a case report of a 8 year old boy with shortening of limbs and enlargement of head. Diagnosis is by physical examination and radiographic documentation.

Keywords : Achondroplasia , disproportionate short stature.

Introduction

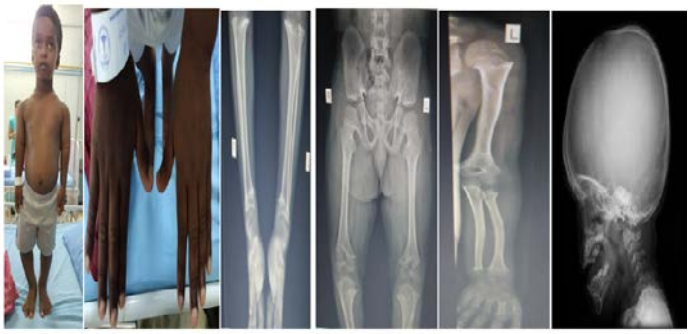
The term Achondroplasia was proposed by Jules Parrott in 1878. In 1900 Pierre marie further investigated and coined term " Trident shaped hand ". Achondroplasia literally translates as without cartilage formation . It is inherited as autosomal dominant trait with complete penetrance, occurs as a result of mutations in one copy of the fibro-blast growth factor receptor 3 gene (FGFR3). The characteristics of achondroplasia are short stature with disproportionately shorter proximal limb bones, narrow trunk, and macrocephaly. There is contraction at the base of the skull with a prominent forehead and flattened midface region and short, broad hands with a trident appearance of the fingers. Disproportionate growth between endochondral bone and the underlying organs leads to a number of orthopedic, neurological, respiratory, ear, nose, and throat (ENT), and dental issues .Whilst serious complications such as sudden death due to severe compression of the spinal cord at the foramen magnum impact on only 5%–10% of children, early monitoring and judicious medical and surgical interventions are important for reducing morbidity and mortality.

Case report

A 8 year old boy was brought to pediatric department with chief complains of shortening of limbs and enlargement of head. The child is second born to non-consanguineous parents, born at term by a normal vaginal delivery at hospital ,No history of NICU admissions. No other family member was previously affected in the family. The child was completely immunized. child had history of frequent respiratory infections got treated in the hospital with each episode. The developmental milestones were delayed . Intelligence of the child is normal ,with good school performance.

On Clinical examination revealed height of less than 3rd per-centile and head circumference more than 97th percentile, head appeared large with frontal bossing, depressed nasal bridge, both upper and lower limbs are short and narrow trunk , Hands were broad with trident configuration. Motor system examination showed mild

hypotonia and reduced muscel power (3/5) in all four limbs.



Skull was large with relatively small skull base , Iliac wings were small and squared, with horizontal acetabular roof ,Limb bones were short with metaphyseal cupping and flaring, with irregular growth plates .Hands were broad with short metacarpals and phalanges with trident configuration.

Discussion

Achondroplasia is the most common form of skeletal dysplasia, affecting growth of tubular bones, spine and skull. Achondroplasia is an autosomal dominant disorder with complete penetration. Mutation of fibroblast growth factor receptor 3(FGR3)gene within the region 4p16.3 was reported as cause of Achondroplasia. In heterozygous state Achondroplasia is nonlethal with normal life span and normal intelligence. In homozygous state Achondroplasia is a lethal condition. When both parents are heterozygos with Achondroplasia, there is 25% risk of homozygous Achondroplasia in their offspring. All patients with typical achondroplasia have mutations at FGFR3 codon 380 . In the FGFR3 mutation group, in which the clinical phenotypes range from severe to mild, the severity appears to correlate with the extent to which the receptor is activated. The mutation maps to the transmembrane domain of the receptor is thought to stabilize receptor dimers that enhance receptor signals, the consequences of which inhibit linear bone growth.Characterstics clinical features are typically present at birth with short limbs, a long narrow trunk, and a large head with midfacial hypoplasia and prominent forehead. The limb shortening is greatest in the proximal segments (rhizomelic limb shortening) and the fingers often display a trident configuration. Most joints are hyperextensible, but extension is restricted at the elbow. A thoracolumbar gibbus and Lumber lordosis are often found. Progressive narrowing of interpedicular distance between L1-L5, smaller greater sciatic notch, short tubular bone, square shape illiacs, contracted base skull and long fibula. Usually, birth length is slightly less than normal but occasion-ally plots within the low-normal range . Skeletal radiographs confirm the diagnosis. The calvarial bones are large, whereas the cranial base and facial bones are small. The vertebral pedicles are short throughout the spine as noted on a lateral radiograph. The interpedicular distance, which normally increases from the first to the fifth lumbar vertebrae, decreases in achondroplasia. The iliac bones are short and round, and the acetabular roofs are flat. The tubular bones are short with mildly irregular and flared metaphyses. The fibula is disproportionately long compared with the tibia. Intelligence is normal unless central nervous system complications develop.Clinical diagnosis is definite based on physical features (present even at birth).The couples can be counselled regarding risk of recurrence. Prenatal diagnosis of homozygous Achondroplasia can be made by mutation detection at 10-12 week of gestation as against 16-20 week by ultrasonographic examination. Ultrasonographic examination can detect shortening of long bones only in late pregnancy . Prenatal diagnosis can be provided early in pregnancy by DNA based methods on chorionic villi.In Western countries, a surgical procedure such as osteotomy is often proposed when genu varum is present and persists

during childhood. Osteotomy is a preferred surgical treatment for thoracolumbar kyphosis and lumbar stenosis in patients with achondroplasia . The early experience with surgical limb-lengthening procedures resulted in a high incidence of complications such as pain and infections . But more advanced procedures have recently resulted in a significant increase in patient height over a 24-month period .Spinal canal decompression is one of the most common surgical strategies to treat spinal stenosis in patients with achondroplasia, and it can reduce symptoms of lumbar stenosis .Double leg lengthening surgery has been proposed as an alternative to treat the achondroplasia by restoring the normal ratio of the trunk and lower limbs through extension of the lower limbs . After surgery, the tibia and femur are extended an average of 10 cm.

New treatment options for achondroplasia

Recently, there have been large strides taken in terms of finding a medical (drug) treatment to alleviate some of the medical complications observed in achondroplasia. One medication that is currently undergoing early Phase II clini¬cal trials is C-natriuretic peptide (CNP), which antagonizes the downstream effects of the aberrant FGFR3 signal, and has been shown to normalize bone growth in mouse models of achondroplasia.72 In addition to this, other medications and antibodies are also being developed that may also aid in increasing bone growth and decreasing complications in this condition.

Conclusion

Children and adults with achondroplasia are impacted by a variety of medical issues created by the unique complexities associated with a form of disproportionate short stature. Further research on the changes in medical issues presenting across the lifespan for individuals with achondroplasia is needed to further drive health- and community-based services needs and assist with directing appropriate and timely service provision. Most of those with achondroplasia will have a normal or near normal life expectancy. Overall, average life span is about 10 years less than that of the general population.

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