Threats to Development

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## 1-2 Threats to Development

Achondroplasia is a genetic disorder predominantly characterized by dwarfism, where the growth of long bones is inhibited, leading to a distinctive appearance with shortened limbs. The condition is caused by a mutation in the fibroblast growth factor receptor 3 (FGFR3) gene, which is crucial for bone development and maintenance. The mutation results in an abnormal protein that overly restricts the growth of long bones in the limbs and spine (National Human Genome Research Institute, 2018).

#### **Societal and Environmental Influences**

The incidence of Achondroplasia is heavily influenced by genetic factors; however, societal and environmental aspects also play a significant role in managing the disorder. Advanced paternal age is a well-documented risk factor for new mutations leading to Achondroplasia. As men age, the likelihood of mutations in sperm increases, thereby raising the risk of reproducing children with genetic anomalies (Feldman, 2022, Chapter 3). The aspect underscores the importance of genetic counseling for potential parents, especially older fathers, to understand the risks associated with late-age paternity.

Beyond genetics, societal attitudes towards disabilities shape the living conditions and inclusion of individuals with Achondroplasia. Societies with robust support systems, accessibility laws, and inclusive policies tend to provide these individuals with a better quality of life. Environments that lack these supports may inadvertently increase the challenges faced by those with Achondroplasia, ranging from accessibility issues in public spaces to limited opportunities in schools and workplaces.

## **Post-Birth Interventions**

Several medical and therapeutic interventions are critical after a child is born with Achondroplasia. These include regular monitoring for common complications such as obesity, sleep apnea, and spinal stenosis. Early intervention involving physical therapy can

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help manage and prevent some physical complications associated with the disorder. Surgical interventions may also be required to correct severe physical deformities and alleviate discomfort (Healthline, 2016).

Psychological support is equally important. Children with Achondroplasia often face social stigma and isolation due to physical differences. Support groups and psychological counseling can be critical in building self-esteem and coping mechanisms. Educational efforts must also extend into the general public and schools to foster a more accepting and understanding environment, reducing social stigma and promoting inclusivity.

## **Educational and Societal Awareness**

Education about genetic conditions like Achondroplasia is essential for reducing misconceptions and stigma. Awareness programs can help demystify the condition, highlighting the capabilities rather than limitations of those affected. Schools play a pivotal role in this regard by integrating education about genetic diversity into their curricula, which can promote empathy and understanding from a young age.

## The Role of Genetic Counseling

Genetic counseling provides families with the information needed to understand the implications of genetic disorders like Achondroplasia. It assists families in making informed reproductive choices and preparing for their child's medical and social needs. Additionally, prenatal screening and diagnostics allow for early detection, which can prepare parents and medical professionals to address the needs of the newborn immediately after birth.

## **Policy and Infrastructure**

To improve life for individuals with Achondroplasia, policies must enforce and enhance accessibility in architectural design, transportation, and public services. Infrastructure adaptations, such as customized furniture in schools and workplaces, accessible

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building entrances, and modified public transport facilities, are practical measures that can significantly improve daily experiences.

To sum up, while Achondroplasia is a condition determined by genetic mutations, the quality of life for those affected is heavily influenced by societal, environmental, and medical interventions. Effectively managing the condition requires a multidimensional approach that includes medical treatment, psychological support, and societal integration. By fostering environments that emphasize inclusion and support, societies can significantly enhance the lives of individuals with Achondroplasia, allowing them to lead fulfilling and productive lives despite the challenges posed by their genetic condition. Comprehensive efforts in public education, policy development, and community support are essential to achieving these goals.

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