Presenter Information

Dieuwerke Dijk is a physician and a resident in training to become a physician for people with intellectual disability from Groningen, the Netherlands. Apart from her clinical work, she is a PhD student at the multidisciplinary CHARGE clinic of the University Medical Center Groningen. Together with Conny van Ravenswaaij, Gianni Bocca and Adelita Ranchor she works on her research about growth and puberty in CHARGE syndrome. In 2017 she attended the CHARGE conference in Orlando as a Davenport fellow.

Presentation Abstract

Growth retardation affects 60-72% of children with CHARGE syndrome and is therefore one of the most prominent problems. We systematically searched for studies that presented growth data and reviewed them. In this presentation, we will discuss why it is important to monitor growth and what is currently known about growth in CHARGE syndrome. This will include factors that may influence growth in CHARGE syndrome and possible interventions to prevent or minimize short stature. There will be time to share personal experiences, for questions and discussion.

Learning Objectives

- Why is it important to monitor growth?
- What do we know about growth in CHARGE syndrome?
- What possible interventions can prevent or minimize short stature?
Growth in CHARGE syndrome
14th international CHARGE syndrome conference
Dallas, Texas august 2-5 2019

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Why measure growth

• Why measure growth?
• How to measure growth?
• What is known about growth
• Factors that influence growth
• Multidisciplinary approach
• Future research

Why measure growth

• To identify children
  – who have undernutrition
  – who have a medical condition that affects growth
    • Eg kidney problems or celiac disease

What is known about growth in CHARGE syndrome?

What we did to find out..

We searched for published research about growth in CHARGE syndrome
Results

- The largest decrease in growth velocity takes place the first months after birth.
- Height remains below average until adulthood.
- Weight is below average in childhood but increases in adolescence and adulthood.

Factors that may contribute to growth retardation

- Feeding difficulties
- Critical illness, multiple surgeries and hospital admissions.
- Cardiac malformations
- Scoliosis
- Endocrine problems

Endocrine problems

- Hypogonadotropic hypogonadism = absent or low production of sex hormones.
- Growth hormone deficiency
- Hypothyroidism or hypoparathyroidism

Multidisciplinary approach

- Evaluating therapy for hypogonadotropic hypogonadism in boys with CHARGE syndrome
- Develop growth charts for CHARGE syndrome