



Sunday, August 4, 2019

Breakout Session H45 • 4:00-5:00 pm • Grand Ballroom D/E

Growth in CHARGE Syndrome

Dieuwerke Dijk, MD, University Medical Center Groningen

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

DR Dijk, CMA van Ravenswaaij-Arts, G Bocca
University Medical Center Groningen
The Netherlands



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- 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

How to measure growth

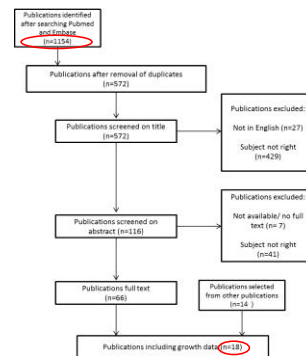
- What is normal growth?
- Based on
 - average growth in a similar group
 - Parental growth



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

Growth-related issue	0-3 years	4-12 years	13-17 years	18+	Professionals involved
Monitoring of growth	Height, weight, head circumference	Height, weight	Height, weight	Weight	Pediatrician General practitioner
Feeding difficulties	Chesnal atresia Cleft lip/palate Cranial nerve abnormalities Vascular ring	Cranial nerve abnormalities Vascular ring Dental problems	Cranial nerve abnormalities Vascular ring Dental problems	Cranial nerve abnormalities Dental problems	Speech and language therapist Pediatrician (pediatric) neurologist Ear nose throat specialist Dentist Plastic surgeon (Pediatric) cardiologist (vascular ring)
Absent feeding behavior	Functional, sensory and psychological evaluation	Functional, sensory and psychological evaluation Think of Sensory problems, intellectual disability, feeding experiences in the past	Functional, sensory and psychological evaluation		Speech and language therapist Pediatrician Psychologist Intellectual disability physician (Netherlands)
Gastro-intestinal problems	Evaluation with special attention to reflux, constipation, abdominal migraine	Evaluation with special attention to reflux, constipation, abdominal migraine	Evaluation with special attention to reflux, constipation, abdominal migraine	Evaluation with special attention to reflux, constipation, abdominal migraine	Pediatrician (pediatric) gastroenterologist Intellectual disability physician (Netherlands)

Multidisciplinary approach

Growth-related issue	0-3 years	4-12 years	13-17 years	18+	Professionals involved
Chronic critical disease					
Cardiovascular problems	Combine surgeries and hospital admissions. Consider enriched diet/tube feeding	Combine surgeries and hospital admissions. Consider enriched diet/tube feeding	Combine surgeries and hospital admissions. Consider enriched diet/tube feeding		Pediatric cardiologist Pediatrician Dietician
Multiple surgeries and/or hospital admissions	Combine surgeries and hospital admissions. Consider enriched diet/tube feeding	Combine surgeries and hospital admissions. Consider enriched diet/tube feeding	Combine surgeries and hospital admissions. Consider enriched diet/tube feeding		Pediatrician Speech and language therapist Dietician
Orthopedic problems					
Scoliosis		Physical examination of spine Radiologic evaluation	Physical examination of spine Radiologic evaluation	Physical examination of spine Radiologic evaluation	Orthopedic surgeon Rehabilitation specialist
Endocrinological problems					
Hypogonadotropic hypogonadism	Before 6 months of age: LH and FSH testing	Small test: LH, FSH, Estrogen/ testosterone testing (from a 11 years of age)	Small test: LH, FSH, Estrogen/ testosterone testing. Start hormone replacement therapy	Continue hormone replacement therapy	(Pediatric) endocrinologist
Hypothyroidism, Hypoparathyroidism	Test when symptoms	Test when symptoms	Test when symptoms	Test when symptoms	Pediatrician (pediatric) endocrinologist
Growth hormone deficiency		Diagnostic tests for GH deficiency. Check for scoliosis	Diagnostic tests for GH deficiency. Check for scoliosis		

Future research

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