

**16th Annual International Meeting**  
**The Velo-Cardio-Facial Syndrome Educational Foundation, Inc.**  
**July 3 – 5, Roma**

## **GROWTH AND FEEDING**

**M.Cristina Digilio**

**Bambino Gesù Pediatric Hospital - Rome**



# Growth and feeding Growth retardation

*J Med Genet* 1997;34:798-804

## Spectrum of clinical features associated with interstitial chromosome 22q11 deletions: a European collaborative study

A K Ryan, J A Goodship, D I Wilson, N Philip, A Levy, H Seidel, S Schuffenhauer, H Oechsler, B Belohradsky, M Prieur, A Aurias, F L Raymond, J Clayton-Smith, E Hatchwell, C McKeown, F A Beemer, B Dallapiccola, G Novelli, J A Hurst, J Ignatius, A J Green, R M Winter, L Bructon, K Brøndum-Nielsen, F Stewart, T Van Essen, M Patton, J Paterson, P J Scambler

DEVELOPMENTAL DISABILITIES  
RESEARCH REVIEWS 14: 3-10 (2008)

## VELO-CARDIO-FACIAL SYNDROME: 30 YEARS OF STUDY

Robert J. Shprintzen\*

Department of Otolaryngology and Communication Science, Velo-Cardio-Facial Syndrome International Center,  
State University of New York Upstate Medical University, Syracuse, New York

40-60 %

*American Journal of Medical Genetics* 45:313-319 (1993)

## Velo-Cardio-Facial Syndrome: A Review of 120 Patients

Rosalie Goldberg, Beth Motzkin, Robert Marion, Peter J. Scambler, and Robert J. Shprintzen  
*The Center for Craniofacial Disorders and the Department of Plastic Surgery (R.G., R.J.S.), the Center for Congenital Disorders and the Department of Pediatrics (B.M., R.M.), Montefiore Medical Center and the Albert Einstein College of Medicine, Bronx, and Blythedale Children's Hospital, Valhalla (R.G., R.M., R.J.S.), New York; Department of Biochemistry and Molecular Genetics, St. Mary's Hospital Medical School, London, United Kingdom (P.J.S.)*

article

January/February 2001 • Vol. 3 • No. 1

## Auxological evaluation in patients with DiGeorge/ velocardiofacial syndrome (deletion 22q11.2 syndrome)

María Cristina Digilio, MD<sup>1</sup>, Bruno Marino, MD<sup>2</sup>, Marco Cappa, MD<sup>3</sup>, Paola Cambiaso, MD<sup>4</sup>, Aldo Giannotti, MD<sup>1</sup>, and Bruno Dallapiccola, MD<sup>5</sup>

## PRESENTING SYMPTOMS AND CLINICAL FEATURES IN 130 PATIENTS WITH THE VELO-CARDIO-FACIAL SYNDROME. THE LEUVEN EXPERIENCE

BY G. VANTRAPPEN<sup>1</sup>, K. DEVRIENDT<sup>2</sup>, A. SWILLEN<sup>2</sup>, N. ROMMEL<sup>2</sup>, A. VOGELS<sup>2</sup>,  
B. EYSKENS<sup>3</sup>, M. GEWILLIG<sup>3</sup>, L. FEENSTRA<sup>1</sup> AND J.P. FRYNS<sup>2</sup>

## THE PHILADELPHIA STORY: THE 22q11.2 DELETION: REPORT ON 250 PATIENTS

BY D.M. McDONALD-McGINN<sup>1</sup>, R. KIRSCHNER<sup>2</sup>, E. GOLDMUNTZ<sup>3</sup>, K. SULLIVAN<sup>4</sup>,  
P. EICHER<sup>5</sup>, M. GERDES<sup>9</sup>, E. MOSS<sup>9</sup>, C. SOLOT<sup>10</sup>, P. WANG<sup>11</sup>, I. JACOBS<sup>6</sup>,  
S. HANDLER<sup>5</sup>, C. KNIGHTLY<sup>12</sup>, K. HEHER<sup>5</sup>, M. WILSON<sup>5</sup>, J.E. MING<sup>1</sup>, K. GRACE<sup>1</sup>,  
D. DRISCOLL<sup>1</sup>, P. PASQUARIELLO<sup>7</sup>, P. RANDALL<sup>2</sup>, D. LAROSSA<sup>2</sup>, B.S. EMANUEL<sup>1</sup>,  
AND E.H. ZACKAI<sup>1</sup>

## Growth and feeding

### Feeding difficulties (40% of the cases)

*Several different causes*



**congenital  
heart  
defect**



**cleft palate**



**hospitalization**



**dysmotility of the  
digestive tract ...**

**Growth and feeding**  
**Feeding difficulties**

**Prenatal manifestation**  
**POLYHYDRAMNIOS**

- **Polyhydramnios is detectable in 10 % of the patients with del22**
- **The onset is generally in the beginning of the second trimester**
- **Decreased swallowing contributes to polyhydramnios**
- **In many patients presenting with severe polyhydramnios, major feeding problems are observed in the first year of life**

**Vantrappen et al., Genet Couns 1999**

## Growth and feeding

### Growth

#### Birth weight

- Normal weight (between 3rd and 97th centile): 84 %
- Weight < 50th centile: 72 %
- Weight < 3<sup>o</sup> centile: 16 %
  
- Mean birthweight (males – 38th week): 2960 g
- Mean birthweight (females – 38th week): 2760 g

Ryan et al., J Med Genet 1997

**Growth and feeding**

**Feeding difficulties**

**Vomiting**

**Nasal regurgitation**

**Coughing**

**Tendency to choking**



## Growth and feeding

### Feeding difficulties

**Vomiting**  
**Nasal regurgitation**  
**Coughing**  
**Tendency to choking**



**Preferred spoon feeding**  
**Difficulties in cup drinking**  
**Preferred consecutive swallows**  
**without taking a breath**



## Growth and feeding

### Feeding difficulties

Vomiting

Nasal regurgitation

Coughing

Tendency to choking



Preferred spoon feeding

Difficulties in cup drinking

Preferred consecutive swallows  
without taking a breath



**Difficulties in chewing with solids**  
**Accepting only a selected few foods**



## Growth and feeding

### Feeding difficulties

#### CAUSES

- **Anatomic defects:**

cleft palate

vascular anomalies (right aortic arch, aberrant subclavian artery)

congenital heart defect

laryngomalacia

- **Functional defects :**

pharyngeal hypotonia

glossoptosis

gastro-esophageal refluxes

dismotility and hypotonia of the digestive tract

infections of the respiratory tract

Growth and feeding

Feeding difficulties

CAUSES

Gastrointestinal malformations:

- esophageal atresia
- intestinal atresia
- Hirschprung disease
- imperforate anus

*JOURNAL OF MEDICAL GENETICS, February 1999, Vol 36, No 2, p 137-139*

Microdeletion 22q11 and oesophageal atresia

Maria Cristina Digilio, Bruno Marino, Pietro Bagolan, Aldo Giannotti, Bruno Dallapiccola

*J Med Genet* 1997;34:79-82

Anal anomalies: an uncommon feature of velocardiofacial (Shprintzen) syndrome?

S Worthington, A Colley, K Fagan, Kang Dai, A H Lipson\*

**Growth and feeding**

**Feeding difficulties**

**EVALUATION**

- **Endoscopic evaluation**
- **Barium swallow**
- **Angio MRI of the aortic vessels**

**Growth and feeding**

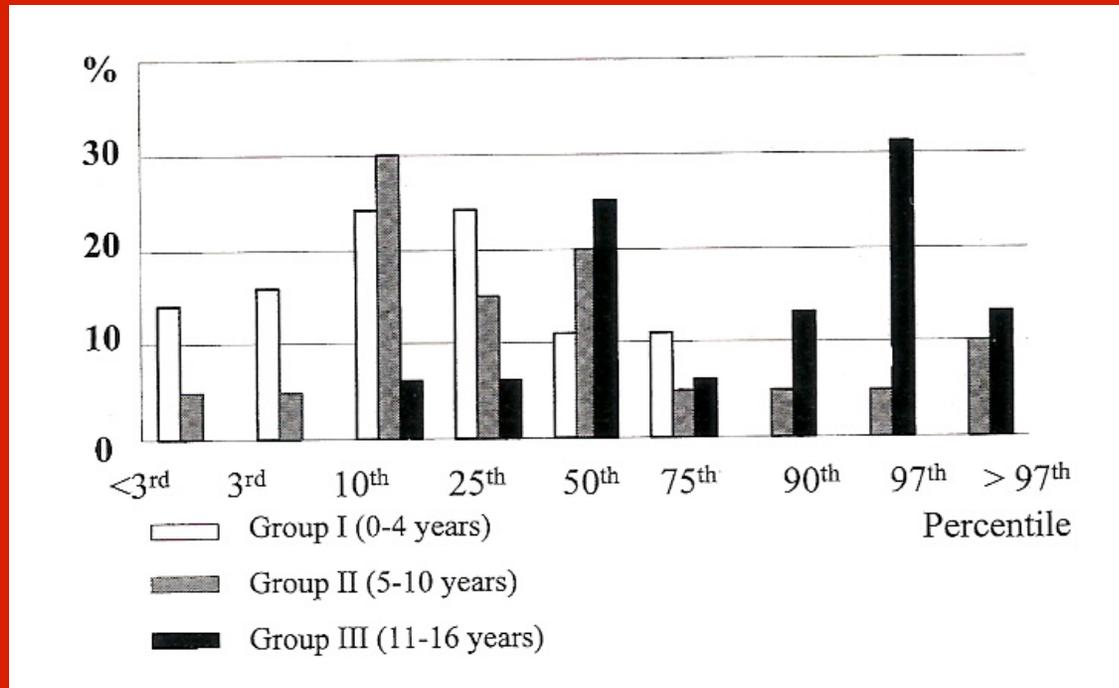
**Feeding difficulties**

**TREATMENT**

- **Feeding techniques**  
(frequent feeding with small quantities, specific nipples, upright position)
- **Medical treatment of gastro-esophageal reflux**
- **Gavage feeding, gastrostomy**
- **Surgical correction of malformations**

## Growth and feeding

### Weight



- Weight deficiency in the first years of age
- Weight normalization in the following years
- Predisposition to develop obesity in adolescence

article

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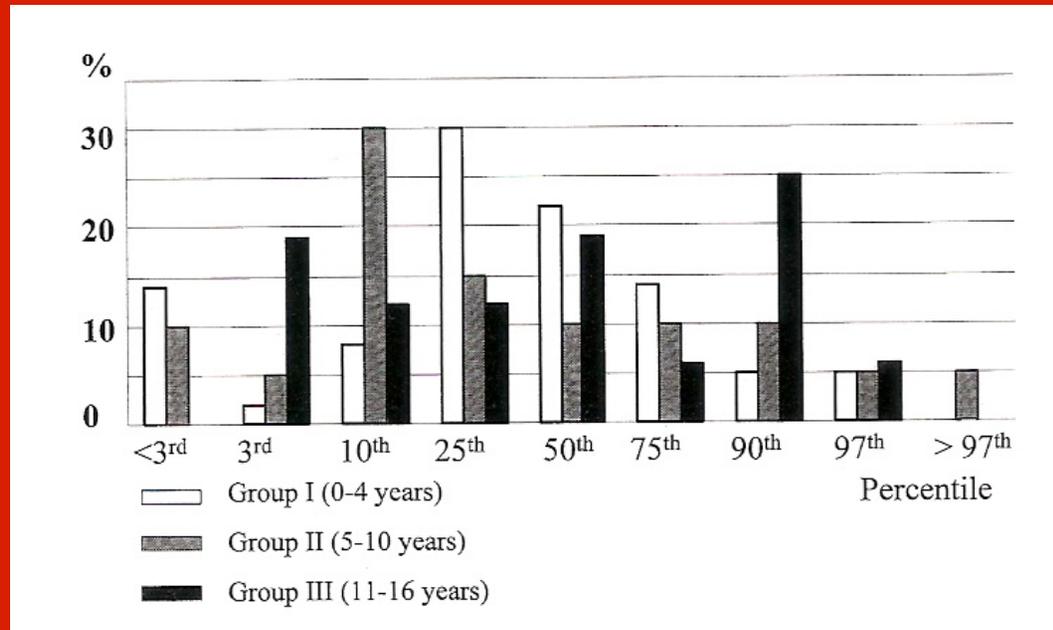
## Growth and feeding

### Causes of weight deficiency

Cause	Comparison	
• CONGENITAL HEART DEFECT	not significant	(p = 0.690)
• CLEFT PALATE	not significant	(p = 0.756)
• FEEDING DIFFICULTIES	significant	(p 0.004)

## Growth and feeding

### Height



- Short stature (<3rd centile) in 14 % of the cases
- The patients with short stature were all < 10 years old
- Normal height in adolescence

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## Growth and feeding



## Bone age

- **1-4 years:** delay in bone age, corresponding to – 4/6 months
- **5-10 years:** delay in bone age, corresponding to – 4/6 months
- **11-16 years:** bone age corresponding to chronological age

## **Growth and feeding**

### **Growth hormone deficiency**

- **Rare**
- **Small pituitary gland**

## Growth and feeding

### Celiac disease

- The prevalence of celiac disease (CD) in patients with Del22 (2%) is not so high as in other types of genetic syndromes
- Screening for CD is indicated only in subjects presenting with persistent gastrointestinal symptoms or significant growth abnormalities

American Journal of Medical Genetics 121A:286-288 (2003)

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*Research Letter*

**Screening for Celiac Disease  
in Patients With Deletion 22q11.2  
(DiGeorge/Velo-Cardio-Facial Syndrome)**

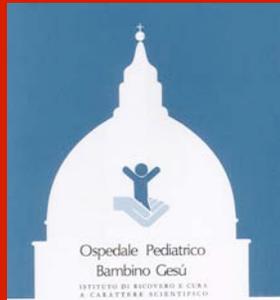
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**Digilio MC, Giannotti A, Castro M, Colistro F,  
Ferretti F, Marino B, Dallapiccola B.**

## **Growth and feeding**

### **Growth: Periodical evaluations**

- **Evaluation of growth parameters (weight and length/height)**
- **Bone age every 2 years**
- **IGF1 blood dosage and pharmacological stimulation of growth hormone (GH) response in patients with height below the 3rd centile**
- **Annual evaluation of blood thyroid hormones (TSH, FT3, FT4) and, after 10 years of age, dosage of antiperossidasis antibodies (AbTPO)**



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