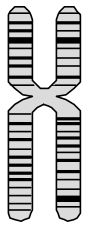


JANUARY, 2004

# ADAMS COUNTY CHILD HEALTH NOTES

Promoting early identification and partnerships between families, primary health care providers & the community.

Distributed by: Adams County Interagency Coordinating Council and Adams County Health Department  
Contributors: Washington State Department of Health and UW – Center on Human Development & Disability



## VELO-CARDIO-FACIAL SYNDROME (VCFS)

### WHAT IS VCFS?

This syndrome was first known as Shprintzen syndrome (recognized in 1978 by Robert Shprintzen, PhD) and is now known to include DiGeorge syndrome at the severe end of the spectrum. VCFS is associated with a deletion of a small segment of the long arm of chromosome 22, hence the regrettable nickname “Catch 22.” Most cases arise from a new mutation, but in 10-15%, transmission is autosomal dominant. Estimated prevalence is 1:2000 people.

*“It was very helpful when the nurse practitioner in our doctor’s office took the time to get us information off the Internet about VCFS... She made us feel that we weren’t just another number in the system. When we first got the diagnosis, we were all alone, we just wanted to talk to SOMEONE. It was great to talk to a parent from the VCFS Network.”*

Parent of a child with VCFS

### WHAT FACTORS SHOULD MAKE YOU CONSIDER VCFS?

VCFS is extraordinarily variable. No single feature is invariant.

Primary defects are of the *velum* (palate), heart and face.

- ✗ Cleft palate (including incomplete or sub-mucous, rarely cleft lip), or merely poor tone of the pharyngeal muscles, potentially leading to swallowing problems and/or otitis.
- ✗ Cardiac defects include ventricular septal defect, right aortic arch, tetralogy of Fallot, and aberrant left subclavian artery. Some children may have no cardiac problems.
- ✗ Craniofacial defects may not be obvious in the early years of life and may include long, prominent nose with squared nasal root and narrow alar base, narrow palpebral fissures, abundant scalp hair, and a “long” face with retruded mandible.

Other common features may include:

- ✗ Delayed speech development. Speech often hypernasal from hypotonic pharyngeal musculature or from cleft palate.
- ✗ Dental enamel hypoplasia
- ✗ GE Reflux, nasal regurgitation of feeds
- ✗ Hypotonia with hyperextensible hands and fingers
- ✗ Mild intellectual impairment in 50%; learning disabilities (mild to severe)
- ✗ Poor growth (either due to inadequate nutrition or to intrinsic growth deficiency associated with VCFS)
- ✗ Visual problems such as far-sightedness, iris deficiencies, small optic discs, tortuous retinal vessels

### WHAT MIGHT YOU DO IN YOUR OFFICE PRACTICE?

- ? Monitor growth (ht, wt, OFC) as plotted on a growth chart at each visit.
- ? Monitor hearing if velum abnormalities are present; refer to audiologist if hearing concerns.
- ? Refer to a genetics clinic for evaluation of the child and parents.
- ? Refer to a craniofacial clinic for treatment related to cranio-facial issues.
- ? Refer for speech therapy, physical therapy, occupational therapy, educational assistance as indicated.
- ? Refer family to local resources/support groups. Families often feel isolated when a family member is diagnosed with this only recently recognized syndrome.

**Washington Support Network:**

VCFS/DiGeorge Family Support Network of Washington  
Ronda McElroy, Statewide Coordinator

1-888-259-VCFS  
email: rhm10@juno.com

**Local Contact Person:**

**Callie Moore – Adams County Health Department**  
*Infant Toddler Early Intervention Program*  
*Children With Special Health Care Needs Program*

**509-659-3317**

**Seattle:**

Polly Parson  
VCFS Support Network of WA

(206) 523-0142  
[grosbeak@ix.netcom.com](mailto:grosbeak@ix.netcom.com)

**Snohomish County:**

Debbie Riebe  
VCFS Support Network of WA

(425) 290-9722  
[deb2100@hotmail.com](mailto:deb2100@hotmail.com)

**Port Angeles:**

Lane Richards  
VCFS Support Network of WA

(360) 874-2921  
[lealc@tenforward.com](mailto:lealc@tenforward.com)

**Yakima:**

Linda King  
VCFS Support Network of WA

(509) 874-2921  
[LLKING47@hotmail.com](mailto:LLKING47@hotmail.com)

**Spokane:**

Bob & Nickki Woods  
VCFS Support Network of WA

509-326-5458  
[rwoods1@uswest.net](mailto:rwoods1@uswest.net)

**REGIONAL RESOURCES/SPECIALTY CLINICS**

VCFS Support Network of WA  
Parent-to-Parent Support Programs of WA  
WA State Father's Network

[www.snonet.org/vcfs/](http://www.snonet.org/vcfs/)  
1 (800) 821-5927  
[www.fathersnetwork.org](http://www.fathersnetwork.org)  
425-747-4004 ext. 218

Children's Hospital & Regional Medical Center Seattle  
Cranio-Facial Clinic  
Genetics Clinic  
Ear, Nose and Throat (ENT) Clinic

1-877-528-2700  
206-526-2528  
206-528-2665  
206-526-2105

**NATIONAL RESOURCES AND WEBSITES**

VCFS Educational Foundation, Inc.  
Dr. Robert Shprintzen, Executive Director  
Syracuse, NY; <http://www.vcfsef.org/>  
315-464-6590; email: [vcfsef@hscsyr.edu](mailto:vcfsef@hscsyr.edu)

Clinical Genetics Center  
Children's Hospital of Philadelphia  
(215)590-2920  
Quarterly Newsletter, 22q and You

***Please note: Adams County Health District became Adams County Health Department on January 1, 2004. There will be no change in the Infant Toddler Early Intervention Program or the Children With Special Health Care Needs Program at this time as a result of this change. The contact people for these programs continue to be Callie Moore at 659-3317 and Melva Zavala at 488-5256. You can also contact the health department office in Othello to leave messages for Callie at 488-2031.***