

Child Health Notes

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

Distributed by: Spokane Regional Health District - Children with Special Health Care Needs Program. Contributors: Washington State Department of Health and University of Washington - Center on Human Development and Disability. Electronic versions of Spokane Child Health Notes: www.srhd.org/health/women-children/childhealthnotes.asp



"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 child with VCFS

Velo-Cardio-Facial Syndrome (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, also known as 22q11 deletion. Most cases arise from a new mutation, but in 10-15%, transmission is autosomal dominant. Estimated prevalence is 1:2000 people. Many of the findings in infants and children with VCFS are subtle, and the diagnosis may not be suspected at all – leading to delay in treatment for the hypernasal speech (speech that sounds like it is coming through the nose). If VCFS is suspected, chromosome analysis using the FISH study (fluorescence in situ hybridization) can confirm the diagnosis. Early diagnosis is important so that medical treatments and early intervention therapies can begin as early as possible.

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 is 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 you can do in your office:

- ♦ Monitor growth (height, weight, OFC) as plotted on a growth chart at each visit.
- ♦ Monitor hearing if abnormalities of the velum are present; refer to audiologist if there are hearing concerns.
- ♦ Refer to the genetics clinic for evaluation of the child and parents.
- ♦ Refer to the Maxillofacial Program for team evaluation related to cleft palate, hypernasal speech, and other 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.

Velocardiofacial (VCFS) Resources:

National Resources:

VCFS Educational Foundation, Inc. www.vcfsef.org or 1-866-VCFSEF5
Clinical Genetics Center, Children's Hospital of Philadelphia www.cbil.upenn.edu/VCFS/

Local Resources:

Inland Northwest Genetics Clinic (509) 535-2278
Eastern Washington (Region 1) Maxillofacial Program (509) 324-1652
Children with Special Health Care Needs Program
Region 1 counties include: Adams, Asotin, Columbia, Garfield, Ferry, Lincoln, Okanogan, Pend Orielle, Spokane, Stevens, and Whitman. For Washington State families from other counties, call (360) 236-3491 for referral to the appropriate Maxillofacial Program in their area.
Pediatric Audiologists:
Holy Family Hospital Speech & Hearing Center (509) 482-2193
Rockwood Clinic Audiology (509) 838-2531
Spokane Audiology Clinic (509) 835-5111
Spokane Ear, Nose & Throat Clinic (509) 624-2326
Spokane Valley Ear, Nose & Throat Clinic (509) 928-7272
Spokane Parent to Parent Support Program (509) 328-6326
Spokane Smiles Family Support website www.spokanesmilesonline.com
Infant Toddler Network (509) 324-1651



FREE Developmental Screenings

Available for infants and toddlers
under three years of age living in Spokane County

Developmental specialists will screen a child's ability to move, communicate, learn, relate to others, see, hear, and eat.

**New for 2007:
Screenings will be available
most months beginning in January**

Call the Spokane Regional Health District's
Infant Toddler Network at 324-1651
to schedule a **FREE** appointment



Children with Special Health Care Needs
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We're Online!!
www.srhd.org