

The Same Name Campaign

The Int'l 22q11.2 Deletion Syndrome Foundation
www.22q.org



22q11.2 Deletion Syndrome

also known as:

- Velo-cardio-facial syndrome (aka Shprintzen syndrome)
Characteristic face, cardiac, palatal, learning issues
- DiGeorge Syndrome
Absent thymus, hypocalcemia, cardiac defects
- Conotruncal Anomaly Face Syndrome plus others.....

22q11.2 Deletion Syndrome

- VCFS, DGS, etc. - older terms described specific clinical phenotypes prior to discovery of the 22q11.2 microdeletion
- Genetically, no detectable difference in microdeletions found in people with VCFS versus those with DGS or other related syndromes
- Technically, the terms are not synonymous: people with VCFS do not have DGS, even though both have the same underlying 22q11.2 deletion!

The Issue

- Several different names used for the same condition
- Creates confusion among families and professionals
- Families, professionals may not access all available information and resources
- Further divides resources for an already rare syndrome, hampering a unified effort toward research and support

The Campaign

- Foundation recognized need to address name issue in order to move forward with its mission of detection, care, and cure
- *Same Name Campaign*: educational effort aimed at professionals and families, with the goal of establishing consistent terminology for the 22q11.2 deletion syndrome
- Intent is to increase collaboration among groups, with all on the same page, moving in the same direction, supporting the worldwide 22q community

The Campaign

- Foundation is sensitive to concerns of families and other groups who may find it difficult to switch to a different name
- Primary focus on professionals who make initial diagnosis: over time, with more consistent use of the 22q11.2 designation, fewer children newly diagnosed with VCFS, DGS, or other outdated terminology

The Campaign

- Presentations about campaign at professional meetings
- Mailings / publications to professionals and families encouraging consistent use of the 22q11.2 deletion nomenclature instead of older names
- Working toward consensus among professional and support organizations throughout the world
- Encouraging publishers of professional journals to adopt consistent standards for naming the syndrome in all published articles

Think 22q!

www.22q.org