Speech Production in 22q11.2... Current Practice and How to Help

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What we do at QCH…

- **Clinical Assessment and Diagnosis** in children with cleft palate, craniofacial conditions and suspected velopharyngeal dysfunction (includes some children with 22q11.2 Deletion Syndrome)
- Where VPD is suspected this will be supported by **Instrumental Assessment (Videofluoroscopy and Nasendoscopy)**
- **Therapy Services** to children with velopharyngeal dysfunction particularly in diagnostic and post-operative phases
- **Consultative support and collaborative care** with other Speech Pathologists and specialists

Queensland Children's Hospital- State wide Service

- Large Geographical area
- Scattered population
- State-wide service delivery
- Cleft / Velopharyngeal Dysfunction Caseload of 1300 active patients
- 87 of these have 22q11.2 Deletion Syndrome where we are monitoring or managing issues related to speech and VPD
- Direct therapy intervention to approx. 25 children at any one time

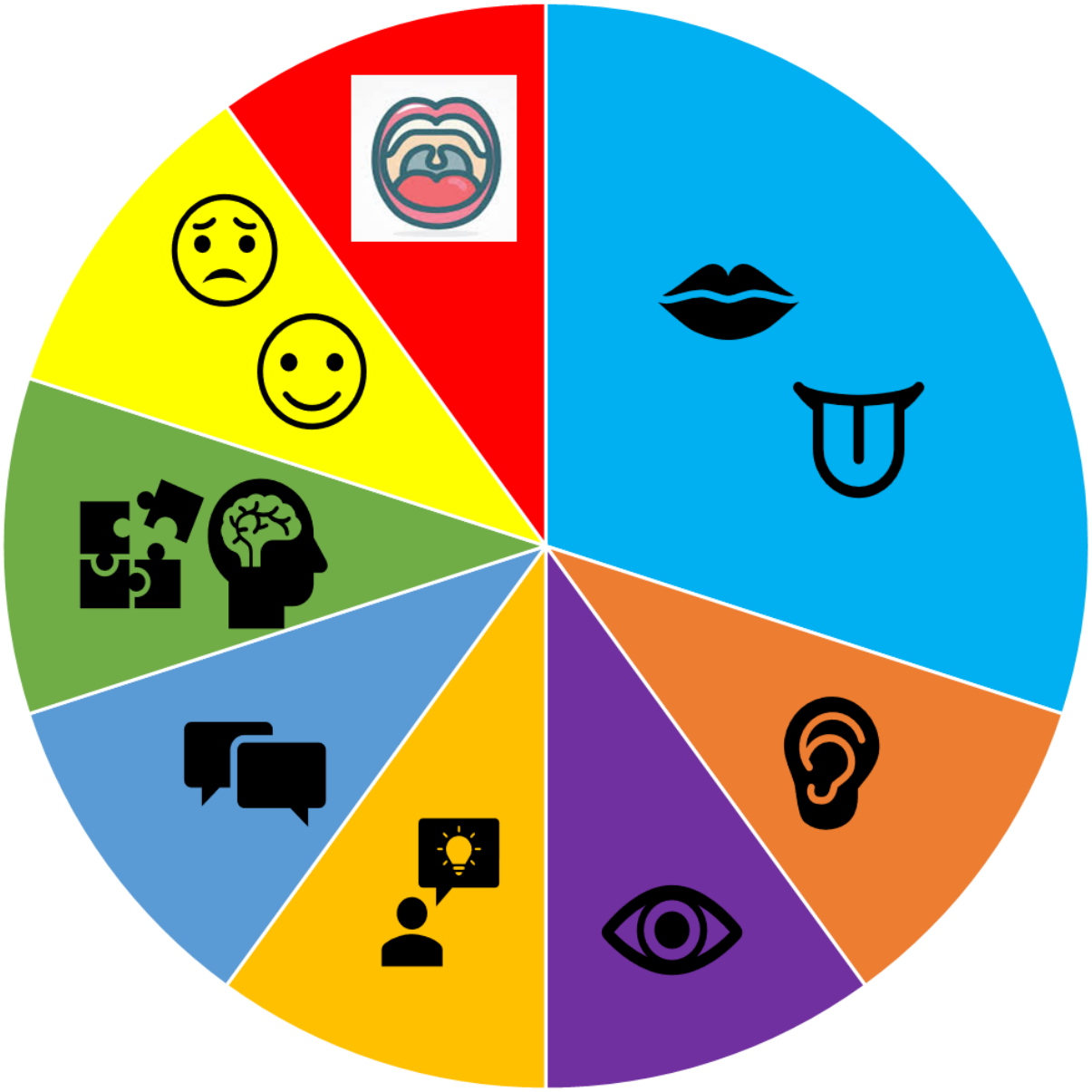
Overview

- Communication Profile in children with 22q11.2 and what influences it?
- What does speech production involve?
- What are the Speech Production Challenges we see in children with 22q11.2
- What is VPD and how do we know if it's a problem
- How is VPD managed if its present?
- What does the evidence say?
- What can we be doing do to help with speech?
- Navigating Services

Developmental Profile in 22q11.2

- Speech Production Difficulties
- Language Delays or Disorders
- Feeding Difficulties
- Psychiatric conditions
- Medical Conditions
- Behavioural Differences
- Learning Difficulties
- Cognitive Issues
- Velopharyngeal Dysfunction and Palate Abnormalities

Speech involves:



- Mouth muscle control - Voluntary movement and co-ordination of mouth muscles
- Hearing - Ability to hear and process speech sounds
- Attention - Ability to attend to spoken information
- Comprehension - ability to understand and follow verbal instructions
- Expressive language development - verbal communication attempts using words
- Cognition / Learning - supports development and consolidation of new skills
- Social participation and emotional / behavioural regulation - support participation in learning expereinces
- Velopharyngeal function - structure and movement of palate muscles to support velopharyngeal closure when correct sound placements are being used

What does the evidence and expert opinion tell us?

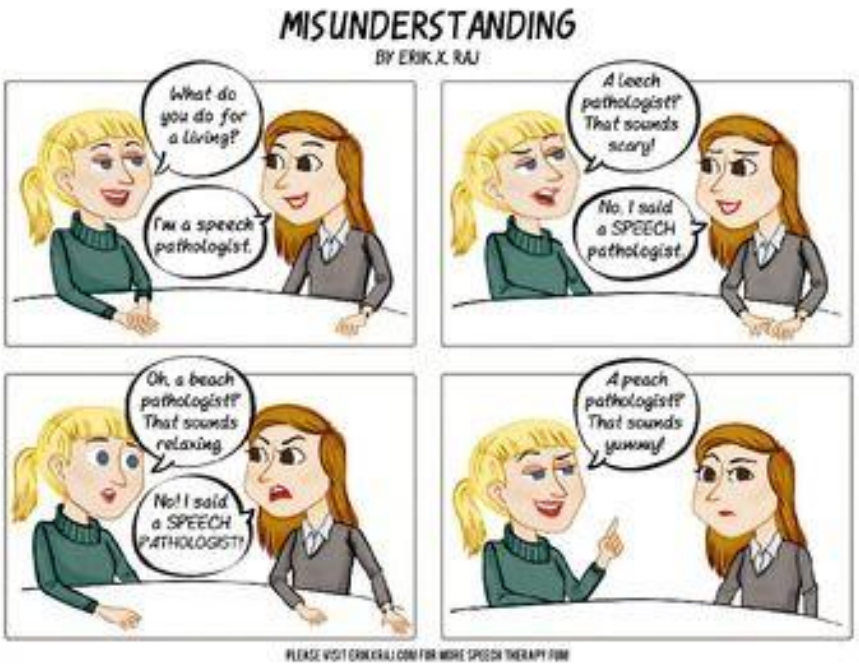
- Disorders of speech and language are a key presenting feature
- Majority of children likely to present with some degree of communication delay and / or disorder

Solot, CB et al. (2019). Speech-Language Disorders in 22q11.2 Deletion Syndrome: Best Practices for Diagnosis and Management. American Journal of Speech-Language Pathology. 28, 984-999

What's the difference?

Speech

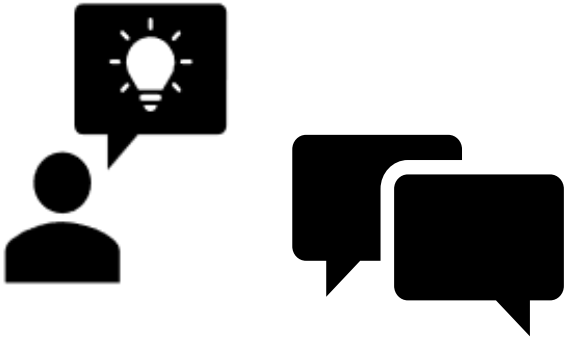
- **The Delivery Platform**
- Its all about how we make and use sounds to communicate the content and meaning



Language

- **The Content and Meaning**
- Expressive and receptive components
- Words
- Putting words together
- Using the right combinations of words in the right situation
- Conveying and understanding what words mean

Language Features in 22q11.2



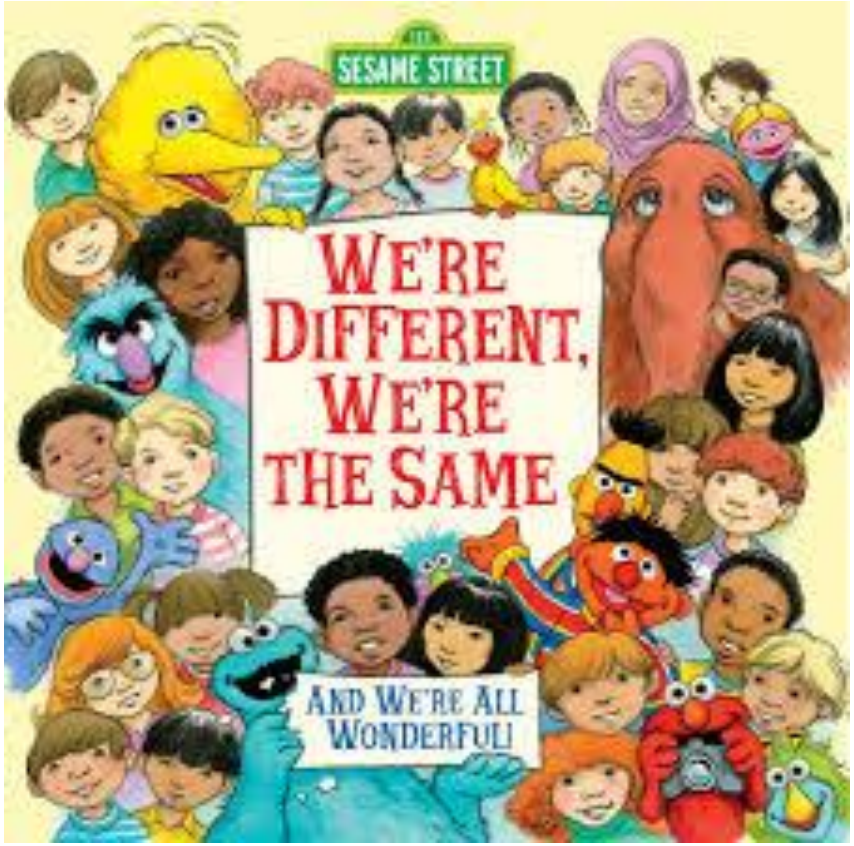
- Receptive and expressive language impairments
- Delayed or reduced babbling and first words
- Slower to start using sentences
- Limited expressive vocabulary
- Delays in phonology (learning of sound rules) may also be present
- Delays in language use e.g. delayed response to questions, reduced sentence length, reduced complexity of language structures
- Difficulties at school age might include: reduced complexity of grammar, difficulties with vocabulary and concept development and abstract reasoning, difficulties with narrative and and descriptive language, possible pragmatic language difficulties.

- Assessment of receptive and expressive language following diagnosis, in preschool years and at school age
- Watch for increase in linguistic demands with age for children who don't display early difficulties

Solot, CB et al. (2019). Speech-Language Disorders in 22q11.2 Deletion Syndrome: Best Practices for Diagnosis and Management. American Journal of Speech-Language Pathology. 28, 984-999

However….

- In spite of a 22q11.2 Deletion diagnosis, every child is unique and needs to be treated based on the issues that present themselves


SESAME STREET
WE'RE DIFFERENT, WE'RE THE SAME
AND WE'RE ALL WONDERFUL!

Making Sense of the Diagnostics??!!!

Phonological Delay or Disorder

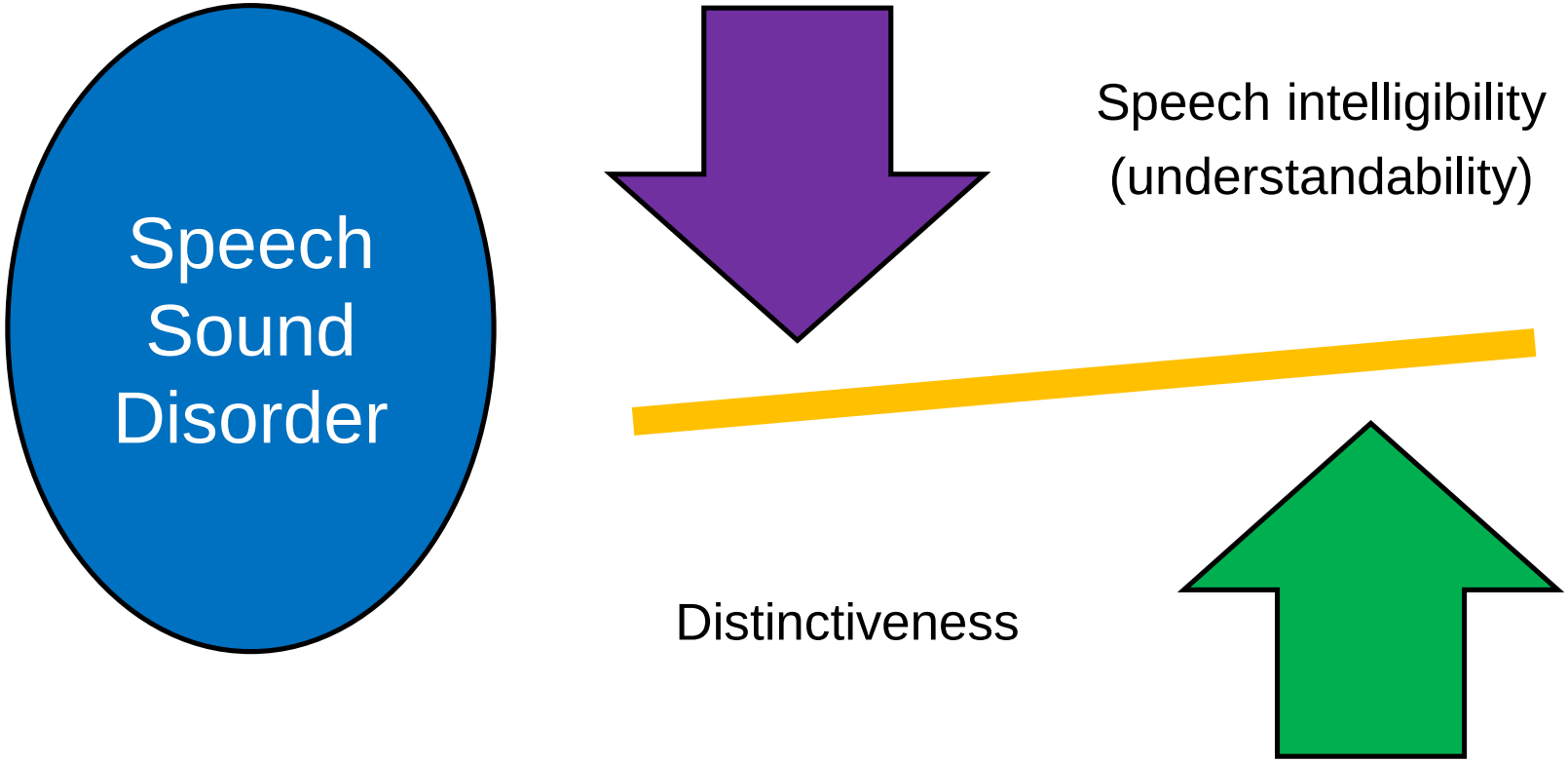
Childhood Apraxia of Speech (CAS)

Speech Production

Dysarthria

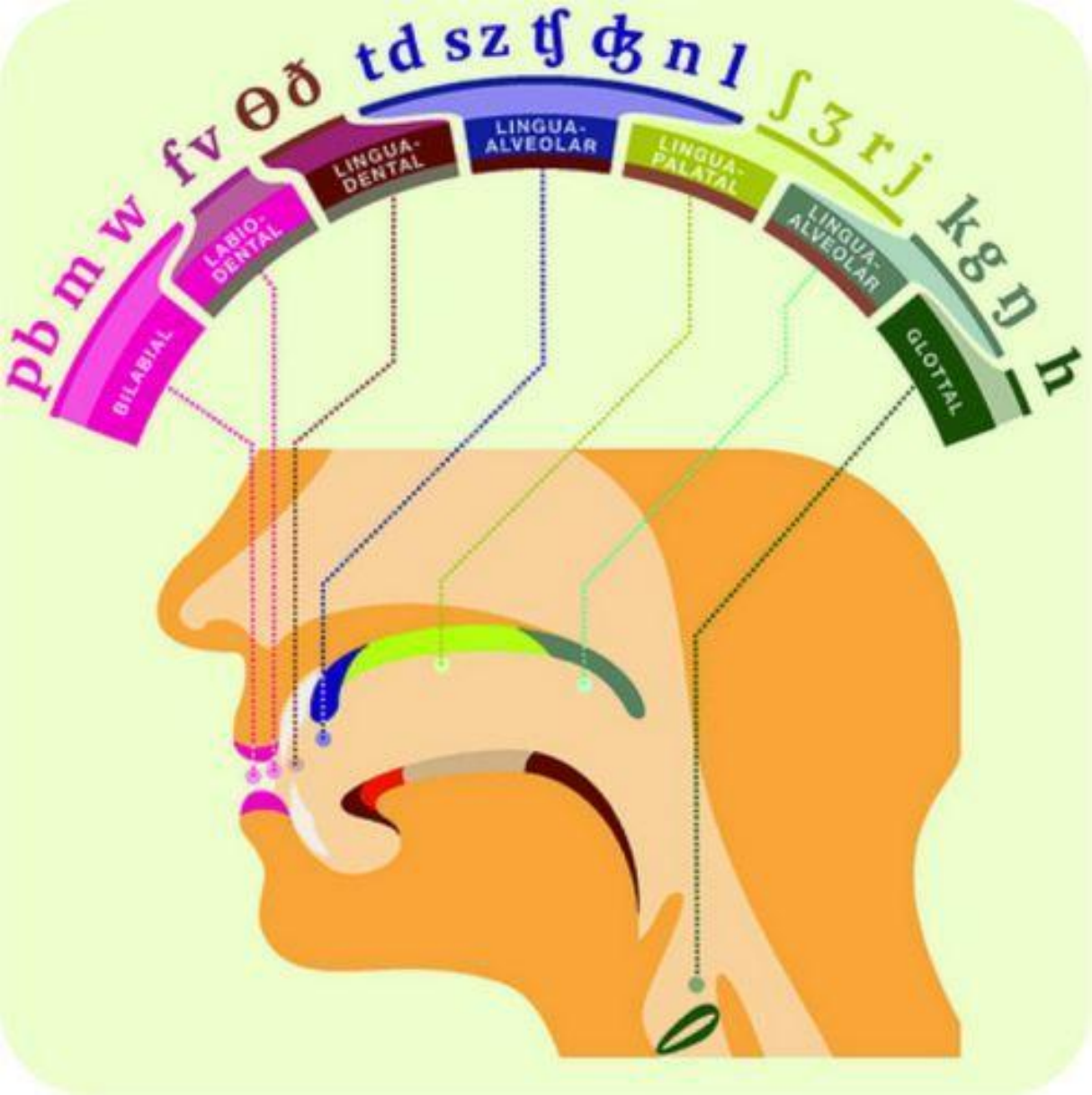
Articulation Based

+/- Velopharyngeal Dysfunction


Speech
Sound
Disorder
Speech intelligibility
(understandability)
Distinctiveness

Articulation Based Errors

- Speech sound production requires movement and contact between the “articulators” – lips, tongue, teeth, gum ridge, hard palate, velum, etc.
- difficulties forming or making individual speech sounds e.g. may lisp / s / sounds like / th /, difficulty making the / r / sound
- may be related to structural differences e.g. missing teeth, bite or jaw relationships


pb m w fv θð td sz tʃ dʒ n l ʃ ʒ r j kg ŋ h
BILABIAL
LABIO-DENTAL
LINGUA-DENTAL
LINGUA-ALVEOLAR
LINGUA-PALATAL
LINGUA-ALVEOLAR
GLOTTAL

Phonological Delay or Disorder

- error patterns often seen as part of normal speech development **OR** may be less typical of normal development

- rule based changes that a child makes when they produce a word (often simplified versions of the word

- may be patterns of:

substitution (replacing one class of sounds for another class of sounds) e.g. child substitutes one sound for another (/ k / to / t /, / f / to / p / etc.)

syllable structure (syllables are reduced, omitted or repeated)
1. cluster reduction (2 or 3 consonants next to each other become one)
2. consonant deletions (e.g. child leaves off the first or last sound in a word)
3. syllable reduction (e.g. child leaves off a syllable/s within the word ("nana" for "banana")

assimilation changes (sounds or syllables start to sound like surrounding sounds) e.g. "lellow' for 'yellow"

Childhood Apraxia of Speech (CAS)

- Difficulty with programming the muscles to do the right thing at the right time

- Problem is at the level of motor speech planning or programming

- Marked by inconsistent errors on consonants and vowels in repeated productions of syllables or words

- lengthened and disrupted articulatory transitions between sounds and syllables

- inappropriate or unusual prosody (unusual word or phrase stress / emphasis)

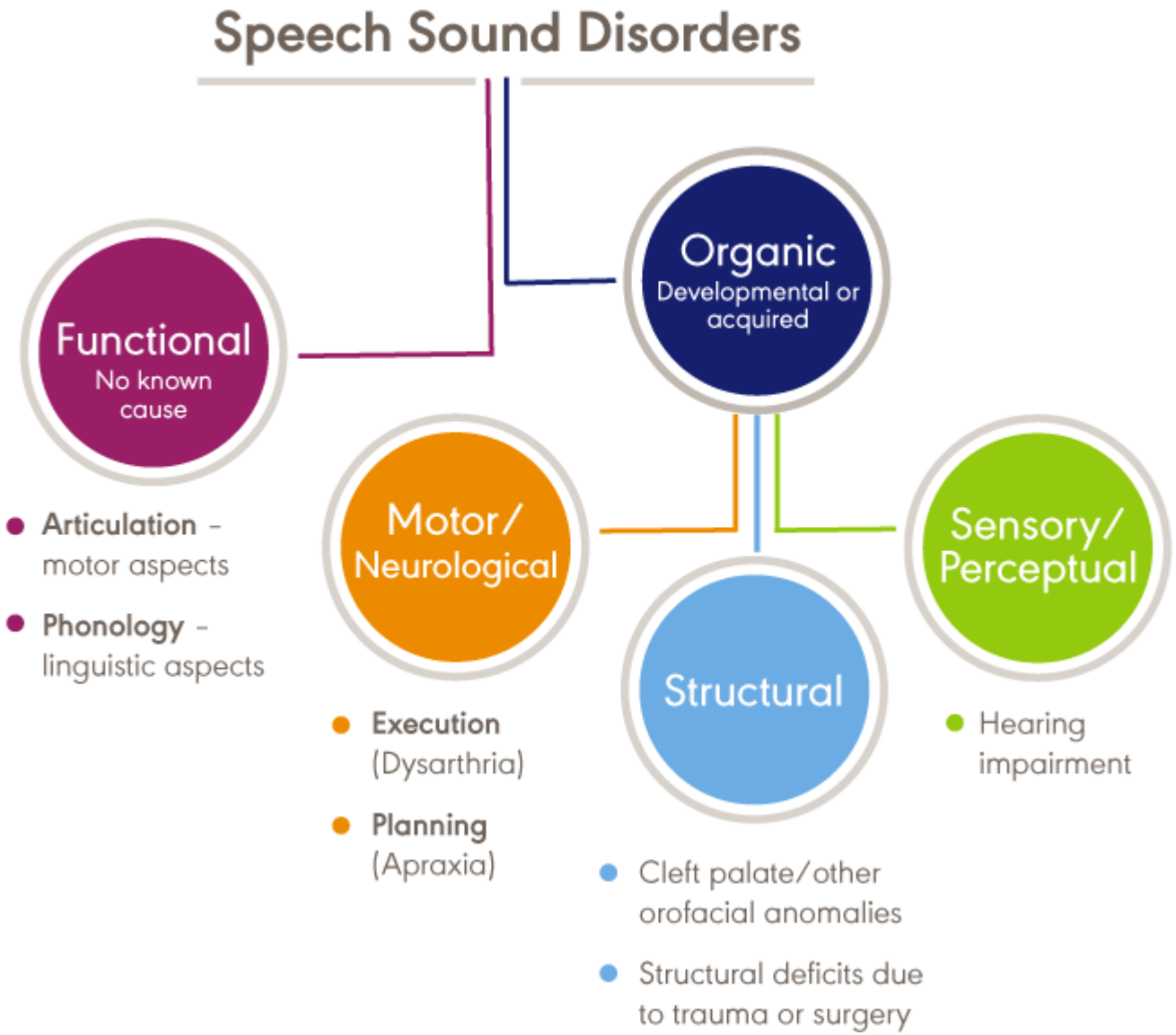
Dysarthria

- Abnormality in strength, speed, range, steadiness, tone or accuracy of muscle movement
- Neuromuscular change affects way speech is produced / execution
- Slower rate of speech
- Indistinct or imprecise speech
- Difficulty controlling volume or loudness of speech
- Differences in voice quality (e.g. breathy, nasal tone)
- Effortful speech due to reduced breath control
- Reduced control of pitch – speech may be monotone, high or low pitched

Speech Motor Delay

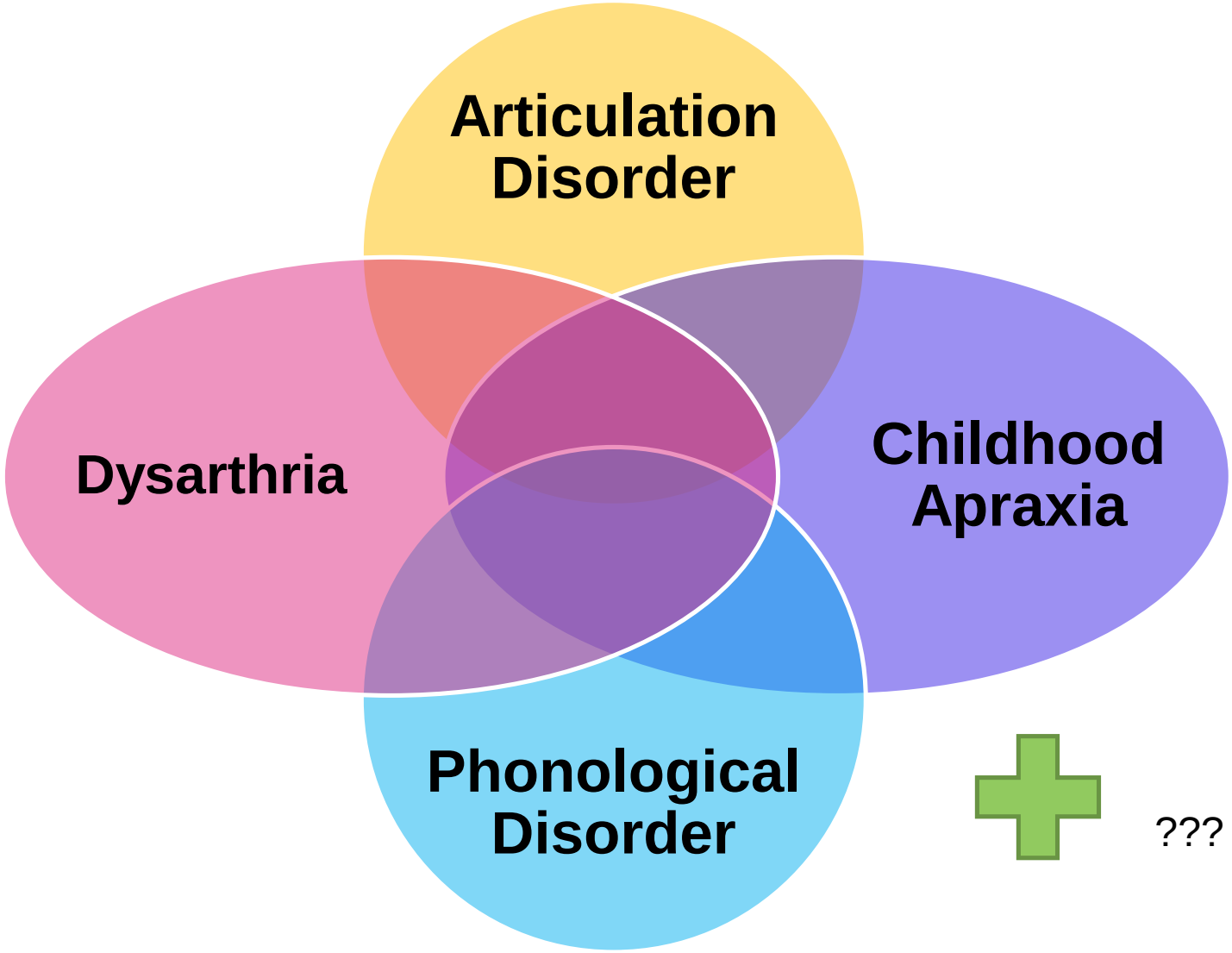
- Newer term being used to describe a delay in speech motor development
- Used for delays in the precision and stability of speech, prosody and voice that do not meet criteria for either childhood dysarthria or CAS

Shriberg, L.D., Kwiatkowski, J., Campbell, T.F., Mabie, H.L., & McGlothlin, J.H. (2018). Motor speech disorders in children with idiopathic speech delay: I Prevalence estimates (Manuscript)



- https://www.asha.org/Practice-Portal/Clinical-Topics/Articulation-and-Phonology/

Unique presentation that can resemble???….



??? VPD

Research on presence of Motor Speech Disorders

- 58.8% of participants with 22q11.2 met criteria for speech delay
- 82.4% met criteria for motor speech disorders
  - 29.4% speech motor delay
  - 29.4% childhood dysarthria
  - 11.8% childhood apraxia of speech
  - 11.8% concurrent childhood dysarthria and CAS

- Motor Speech Disorders were not significantly associated with velopharyngeal dysfunction

Baylis, A.L & Shriberg, L.D. (2018). Estimates of the prevalence of speech and motor speech disorders in youth with 22q11.2 Deletion Syndrome. American Journal of Speech Language Pathology. 1-30.

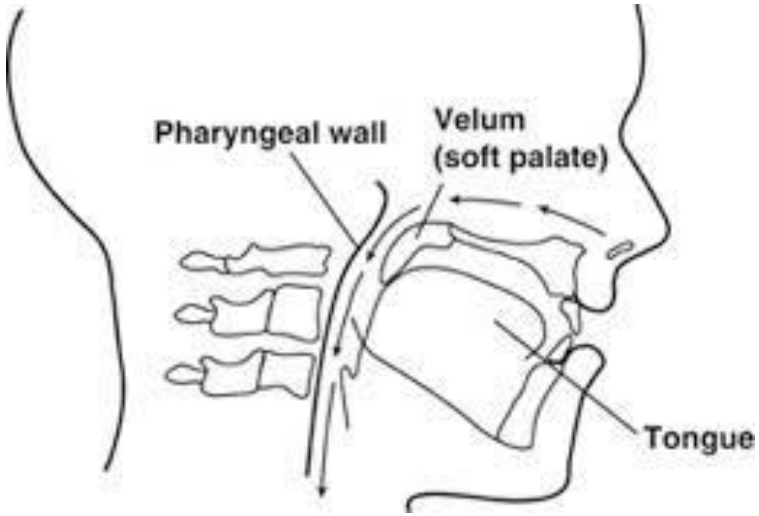
- Speech classifications and motor speech classifications could occur independently of one another in this study

Speech Production features in 22q11.2

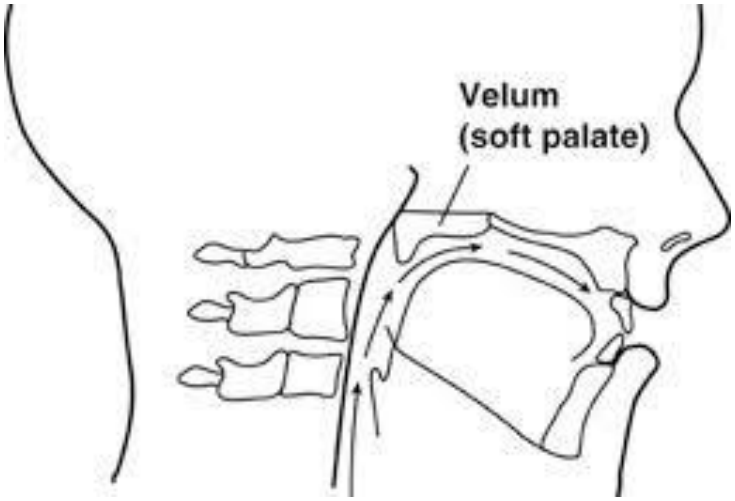
What changes when VPD is in the mix???

What does normal velopharyngeal function involve?

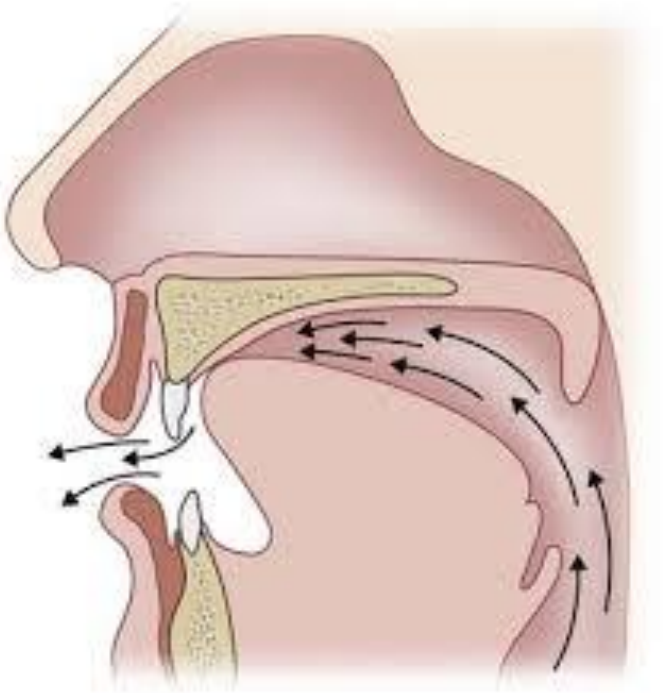
At rest



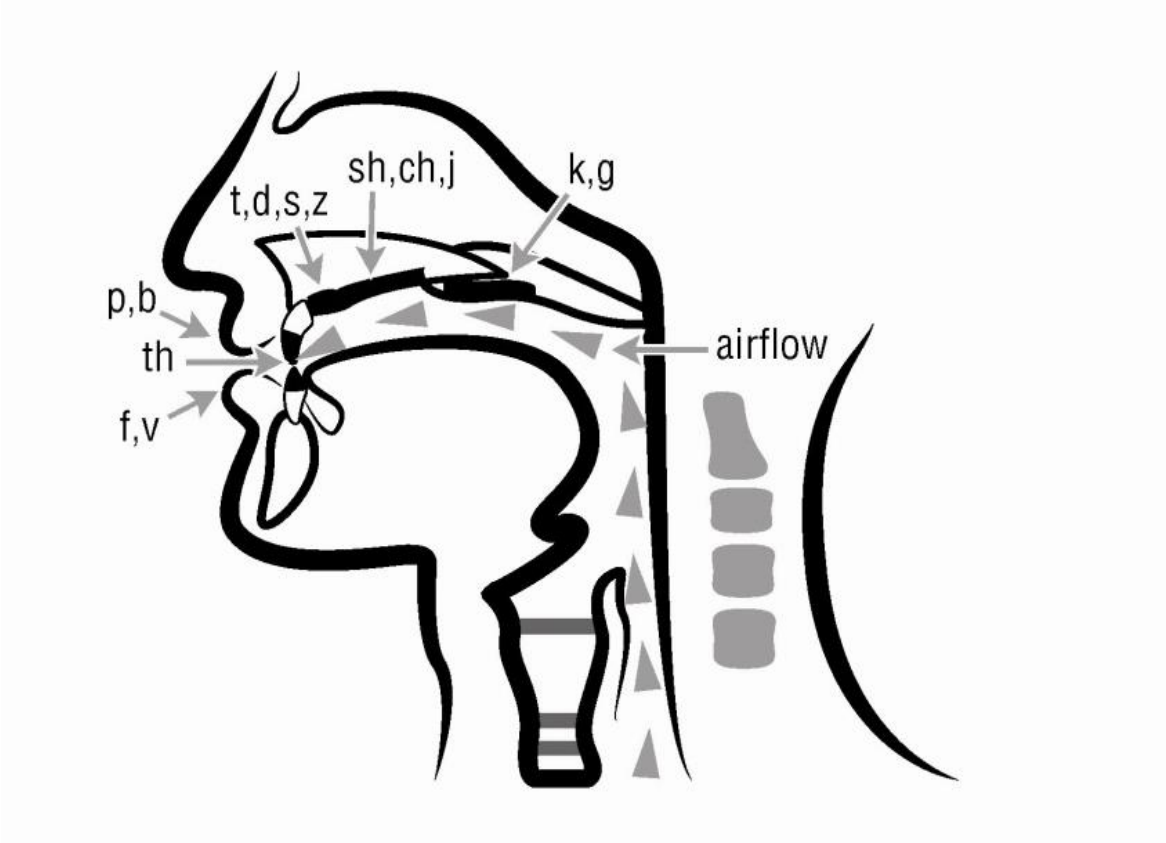
During speech



Normal Velopharyngeal function requires…



- Elevation of soft palate (velum)
- Inward movement of lateral pharyngeal walls
- Anterior movement of posterior pharyngeal wall



- Velopharyngeal closure is required during production of all of the high pressure consonants in speech - / p /, / b /, / t /, / d /, / k /, / g /, / f /, / v /, / s /, / z /, / sh /, / zh /, / ch /, / j / and / th /

What are our clues that there might be problems with VP function?

VPD impacts…

Resonance

Voice = Resonance + Phonation

Nasal Airflow

accompanying pressure consonants

+/- Speech Production

↑ severity = passive features

+/- active errors

Other Characteristics of VPD

- Resonance Changes = **Hypernasality**

  - excessive nasal resonance perceived on vowels and voiced consonants

- Nasal Airflow = **Nasal emission +/- Nasal turbulence**

  - nasal air escape most easily heard on voiceless pressure consonants

- **Nasal or facial grimacing**

  - attempt to reduce nasal airflow by constricting the nares or other facial muscles

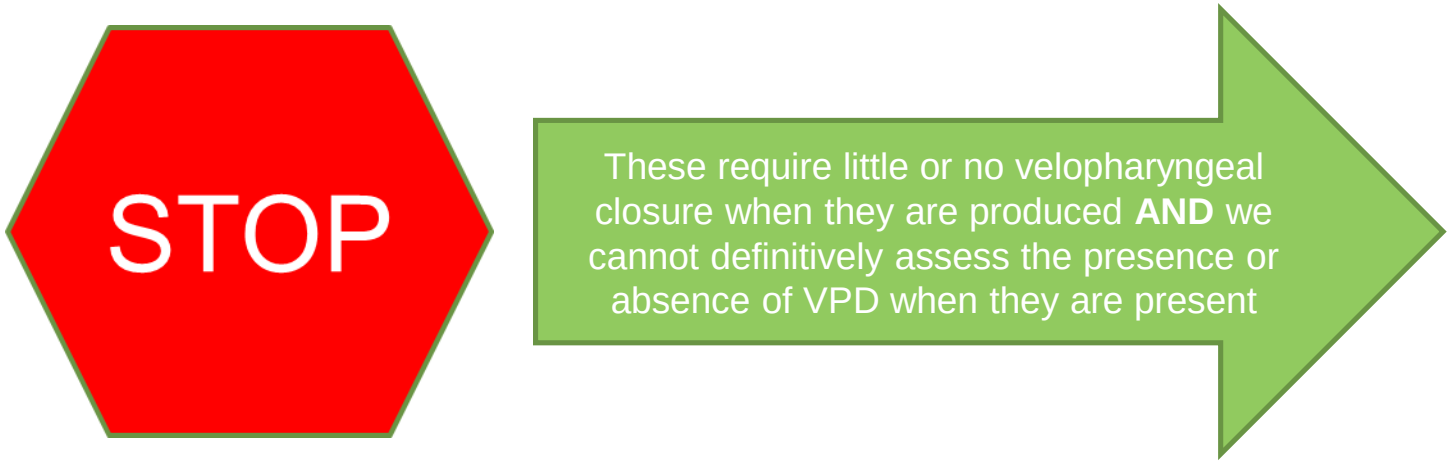
Unique Speech Production Features suggesting that VPD might be present

- child omits all of the high pressure consonant sounds in their words
- May be using only nasal consonants, vowels and low pressure consonant sounds in word attempts
- May make the place of articulation for sounds outside of the mouth ie. **NON-ORAL** speech characteristics
- NON-ORAL speech characteristics are **ACTIVE** and need diagnostic speech therapy first to see what the palate can do.
- These often start as ARTICULATION ERRORS but can become unique PHONOLOGICAL PATTERNS for some children

Speech Characteristics possibly accompanying VPD ???

Non-Oral Speech Characteristics –

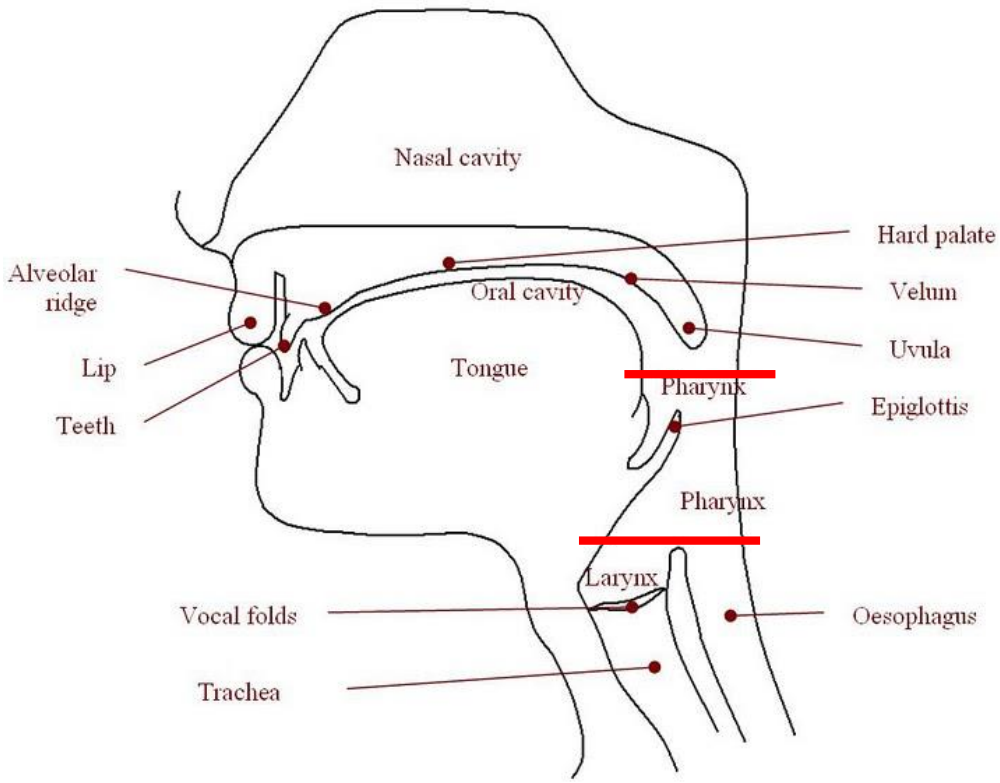
eg. glottal stops, glottal fricatives, pharyngeal fricatives and affricates, active nasal fricatives



Diagnostic

Therapy

Glottal & Pharyngeal Articulations



- Airflow stopped at lower level
- No attempt at VP closure
- Co-articulation common with other articulators
- nose holding

Unique Speech Production Features suggesting that VPD might be present

- **Weak / Nasalized consonants**

  - passive escape of air causes sound to closer to a nasal

  - weak sounding pressure consonants

- **Nasal substitutions for plosive sounds**

  - eg. / b / → / m /, / d / → / n /

- **Passive Nasal Fricatives**

What does VPD look like in children with 22q11?

- Reported VPD incidence rates vary significantly:

  - 74% hypernasal (Rommell, N et al. 1998) N = 50
  - 97% velopharyngeal incompetence assessed by SP or videofluoroscopy or both (Lipson et al. 1991) N = 38
  - 32% velopharyngeal insufficiency (Ryan et al ,1997) N = 558
  - 47% VPI or hypernasal speech (Cohen et al. 1999) N=126 Adults reported in literature

- Reported Syndrome specific findings include:

  - Some VPD related to overt cleft palate, cleft lip and palate or submucous clefts of palate – overt or occult but a number will not have clefts at all
  - 42% VPI; SMCP 16%; CP 11%, CLP 2% (McDonald-McGinn & Sullivan, 2011) N=906

What does VPD look like in children with 22q11.2?

QCH Clinical Group

- 57% being followed or managed for VPD or nasality (N = 153)
- 20% have a pharyngeal flap (N = 87)
- 16% have an overt cleft, cleft types including bilateral cleft lip and palate, unilateral cleft lip and palate, Soft palate cleft, cleft palate, cleft lip
- 19% have nasality without VPD

What does VPD look like in children with 22q11?

- Hypoplasia and hypotonia of the muscles involved in VP closure; wide or deep pharynx; platybasia (obtuse anterior cranial base angle / flatter skull base); cervical spine abnormalities; reduced tonsil and adenoid volume; asymmetrical muscle function; cranial nerve abnormalities; slower or poor timing of VP movement / closure

Solot et. Al (2019); Widdershoven et. Al (2008); Spruijt et al. (2014); McDonald-McGinn & Sullivan (2011)

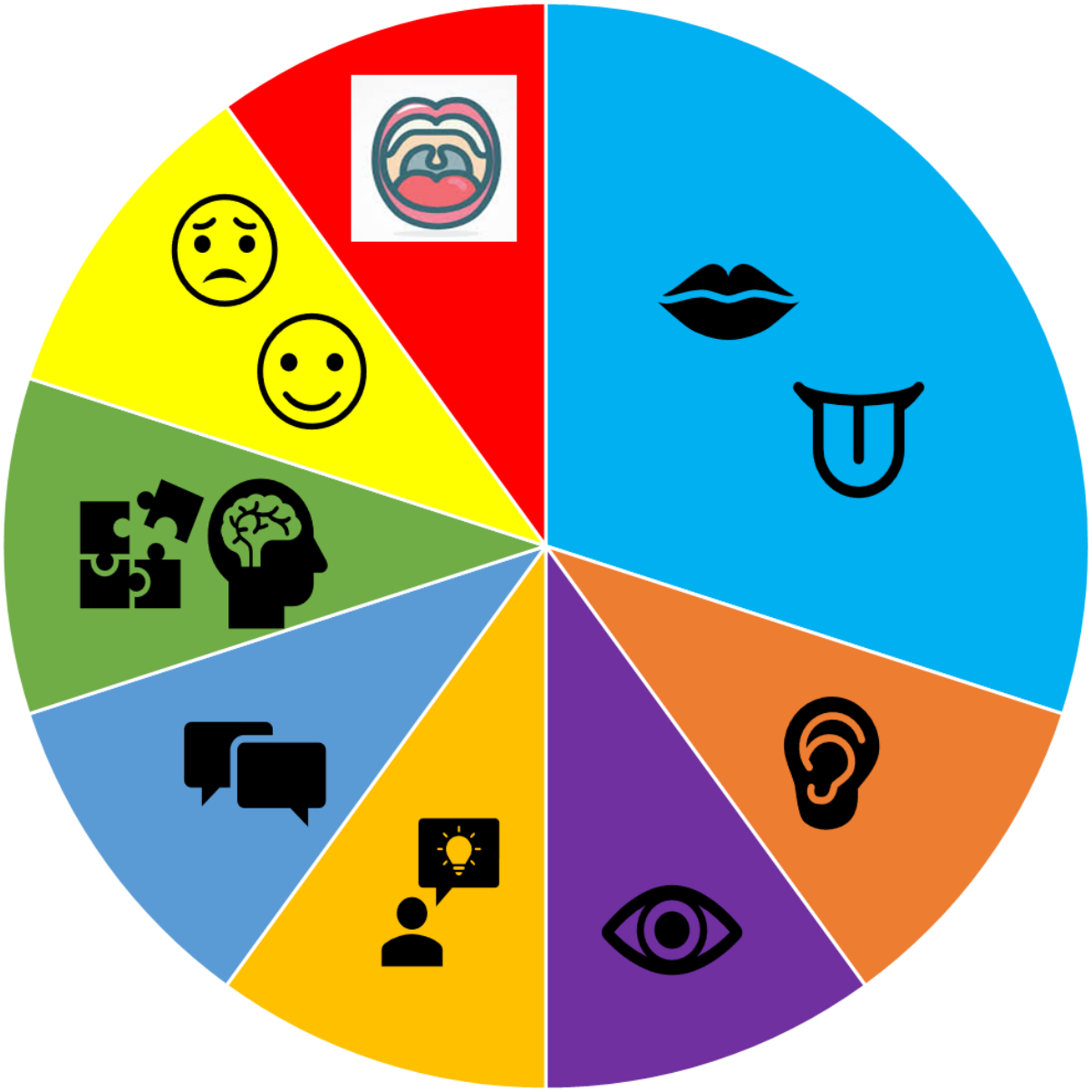
VPD tends to be more severe and persistent than in other patient groups

Jackson et al. (2019); Samoy (2015); Spruijt et al. (2012); Lipson et al. (1991)

Diagnostic Therapy

- Where Non-oral sound productions are present, we need to correct them and establish ORAL (mouth) place of articulation
- Often prior to instrumental assessment to establish oral consonants needed for accurate assessment of velopharyngeal function
- To work on these, we need **language** skills **first**
- Ideally shorter term but can sometimes take a lot longer to establish oral placement for high pressure sounds

Speech involves:



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Don't Bother Doing…

Non-Speech exercises/activity

- palatal exercises
- massage
- blowing
- sucking
- swallowing
- cheek puffing
- Gagging

Ruscello D.M. An examination of nonspeech oral motor exercises for children with velopharyngeal inadequacy. Seminars in Speech and Language, 2008; 29 (4): 294-303.

What do we do about it?

Conduct INSTRUMENTALINVESTIGATION

VIDEOFLUOROSCOPY PALATE STUDY

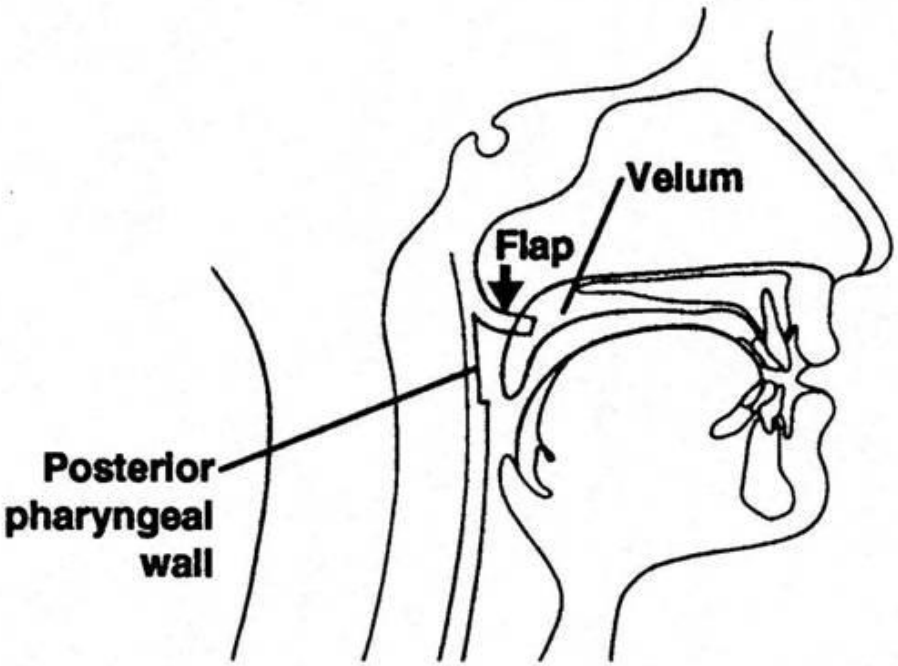
- Younger children > 3 years at earliest
- Need enough language and ability to to produce a speech sample containing high pressure consonant attempts while sitting still in x-ray machine
- Conducted in Medical Imaging
- May require barium contrast into nose

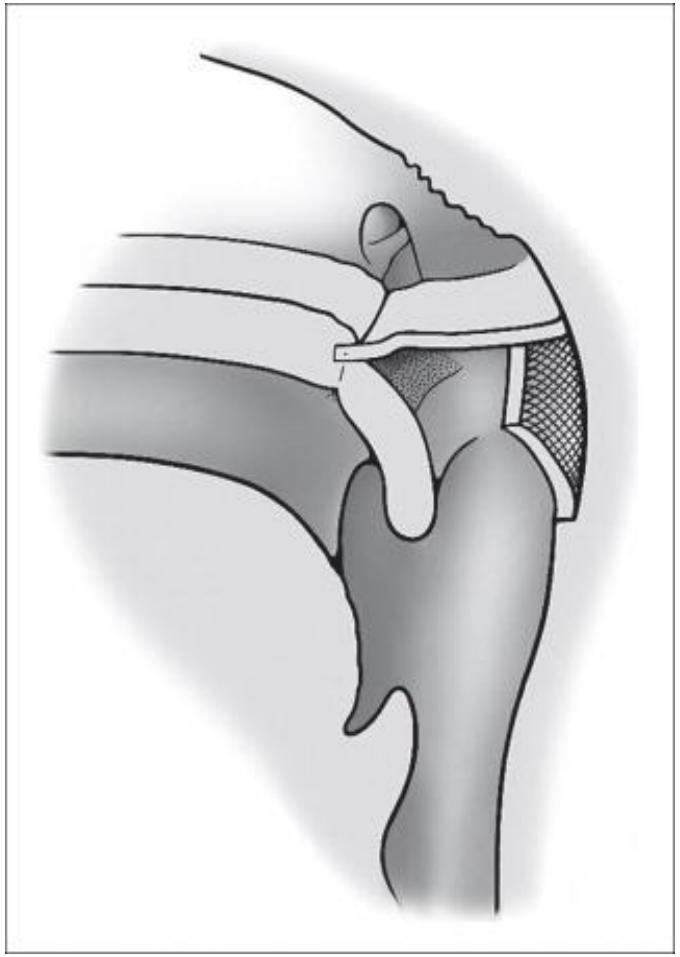
NASENDOSCOPY

- Generally > 5 or 6 years
- Conducted in conjunction with ENT Surgeon
- Requires flexible scope insertion into nose and production of speech sample using high pressure consonant attempts
- Use of topical local anaesthetic

Surgical Management of VPD

Pharyngeal Flap





Surgical Management of VPD

- Remember that the aetiology of VPD in children with 22q11.2 is often multifactorial
- Differential diagnosis of VPD can be very challenging for that reason and can often be delayed
- Diagnosis and management of VPD can be further complicated by the co-occurrence of severe speech sound disorder and/or motor speech deficits
- Motor speech deficits can exacerbate the severity of perceived hypernasality, can limit velopharyngeal closure and /or negatively impact the degree of improvement after VPD surgery

Baylis, A.L & Shriberg, L.D. (2018). Estimates of the prevalence of speech and motor speech disorders in youth with 22q11.2 Deletion Syndrome. American Journal of Speech Language Pathology. 1-30.

Speech Production Features with possible VPD

The interaction between speech production and velopharyngeal function can be a complex one and needs careful diagnosis

- Remember that other speech production issues may also be present
- Speech Pathology's role is to unpack the nature of the speech production issues and other possible VPD impacts
- **Referral on to QCH SP service** indicated in the presence of **NON-ORAL** and **PASSIVE** speech production features AND **RESONANCE and NASAL AIRFLOW differences**

How to help with speech production

- Remember language, listening, attention etc. come first
- Make sure your child can see your mouth and hear you!
- Overemphasise the sounds you want them to try
- Be explicit in telling them what to do and respond positively to all attempts

How to help with speech production

- Changing motor or habitual speech patterns requires repetition and lots of practice but often at a simpler level first

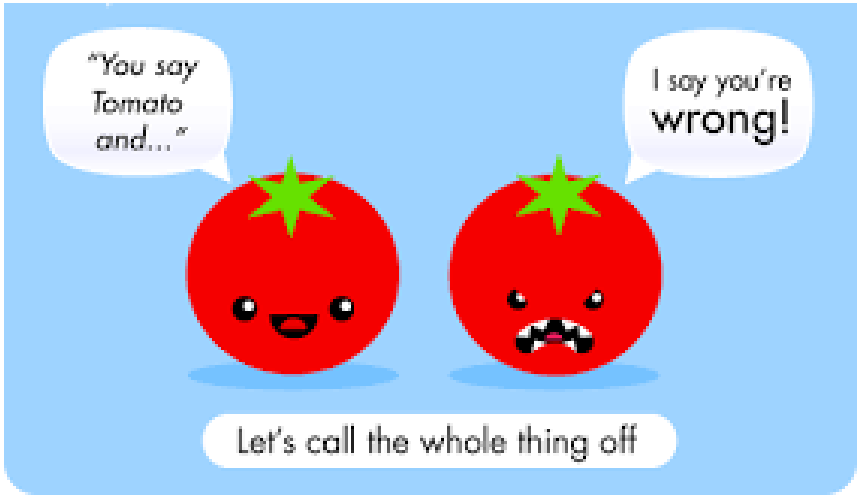
- Isolation → syllables
- Syllables → words
- Words → phrases
- Phrases → sentences

How to help with speech production

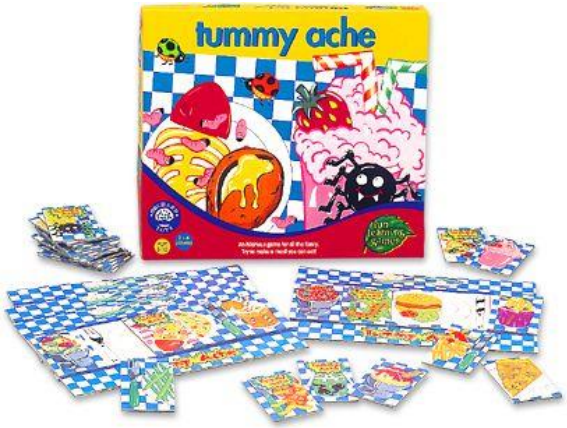
- Try not to correct speech errors all day long, pick a word or two to focus on
- Speech intelligibility or understandability is a priority – pick functional / high use words to practice target sounds. They need to be meaningful to your child!
- Try to get to functional phrases with a target sound
- Show your child how to use the target sound in words in different types of phrases or sentences – this helps language too!

How to help with speech production

- Ask your Speech Pathologist what sounds you should be targeting and what your child's best attempt sounds like
- Always try to notice if your child tries to "self correct"
- Meet your child where they're at, and have fun!
- It's not about perfection, its about function



Keeping it fun and functional….. / p /



What other resources are there?

First 5 Forever

- https://www.slq.qld.gov.au/first-5-forever

Speech Pathology Australia (Fact Sheets)

- https://www.speechpathologyaustralia.org.au/SPAweb/Resources_for_the_Public/Communication_Milestones/SPAweb/Resources_for_the_Public/Communication_Milestones/Communication_Milestones.aspx?hkey=fb6753df-a757-4c4a-8100-aaebdd4451fd

Raising Children

- https://raisingchildren.net.au/disability

Navigating Services

- National Disability Insurance Scheme (NDIS)
- Speech Pathology Australia – Find a Speech Pathologist

https://www.speechpathologyaustralia.org.au/

- Children's Health Queensland - Child Development Services https://www.childrens.health.qld.gov.au/chq/our-services/community-health-services/child-development-program/
- Cleft Palate/Craniofacial/VPD Teams at Australian Children's Hospitals – for VPD concerns – Medical referrals required

References:

Bassett et al. (2011). International 22q11.2 Deletion Syndrome Consortium. (2011). Practical guidelines for managing patients with 22q11.2 deletion syndrome. The Journal of Pediatrics, 159 (2), 332-339.

Bayliss. A.L. & Shriberg L.D. (2018). Estimates of the prevalence of speech and motor speech disorders in youth with 22q11.2 deletion syndrome. American Journal of Speech-Language Pathology. 1-30.

Campbell, et. al (2018). What's new with 22q? An update from the 22q and You Centre at the Children's Hospital of Philedephia. American Journal of Medical Genetics, 176 (10), 2058-2069.

Cohen E. Chow, E.W.C., Weksberg, R. & Bassett, A.S. (1999). Phenotype of adults with 22q11 deletion syndrome: A review. American Journal of Medical Genetics. 86 (4): 359-65.

Dyce O. (2002). Otolaryngological manifestations of the 22q11.2 deletion syndrome. Archives in Otolaryngology Head and Neck Surgery. 128: 1408-12.

Jackson, O.A., Paine, K., Magee, L., Maguire, M.A., Zackai, E., McDonald-McGinn, D.M., McCormack, S. & Solot, C. (2019). Management of velopharyngeal dysfunction in patients with 22q11.2 deletion syndrome. A survey of practice patterns. International Journal of Pediatric Otorhinolaryngology. 116: 43-48.

Lipson, A.H.; Yuille, D., Angel, M., Thompson, P.G., Vandevoord, J.G. & Beckenham, E.J. (1991). Velocardiofacial (Shrintzen) syndrome: An important syndrome for the dysmorphologist to recognise. Journal of Medical Genetics, 28: 596-604.

McDonald McGinn D.M., & Sullivan, K.E. (2011). Chromosome 22q11.2 deletion syndrome (DiGeorge Syndrome/Velocardiofacial Syndrome). Medicine, 90 (1): 1-18.

McDonald-McGinn D.M., Sullivan, K.E., Marino, B., Philip, N., Swillen, A., Vorstman, J.A.S., Zackai, E.H., Emanuel, B.S., Vermeesch. J.R., Morrow, B.E., Scambler, P.J. & Bassett.A.S. (2015). 22q11.2 deletion syndrome. Nature Reviews Disease Primers. 1, 15071: 1-19.

Ryan A.K. et al. (1997). Spectrum of clinical features associated with interstitial chromosome 22q11 deletions. A European collaborative study. Journal of Medical Genetics. 34: 798-804.

Samoy K. et alRommel, N., Vantrappen, G., Devriendt, K., Swillen, A., Feenstra, L., & Fryns, J-P. (1998). Retrospective analysis of feeding and speech disorders in 50 patients with velo-cardio-facial syndrome. Genetic Counselling, 10 (1) 104.

References:

Samoy, K., Hens, G., Verdonck, A., Schoenaers, J., Dormaar, T., Breuls, M. & Vander Poort, V. (2015). Surgery for velopharyngeal insufficiency: The outcomes of the university hospitals Leuven. International Journal of Pediatric Otorhinolaryngology. 79: 2213-2220.

Solot C.B., Sell, D., Mayne, A., Bayliss, A.L., Persson, C., Jackson, O. & McDonald-McGinn, D.M. (2019). Speech-Language disorders in 22q11.2 deletion syndrome: Best practices for diagnosis and management. American Journal of Speech-Language Pathology. 28. 984-999.

Spruijt N.E., ReijmanHinze, J., Hens, G., Vander Porten V. & Van Der Molen, A.B.M. (2012). In search of the optimal surgical treatment for velopharyngeal dysfunction in 22q11.2 deletion syndrome: A systematic review. PLoS ONE 7 (3): e 34332 doi:10.1371/journal.pone.0034332.

Spruijt, N.E., Kon, M., Van Der Molen, A.B.M. (2014). Platybasia in 22q11.2 Deletion syndrome is not correslated with speech resonance. Archives of Plastic Surgery. 41: 344-349.

Spruijt, N.E., Vorstman, J.A.S., Kon, M., VanDer Molen, A.B.M. (2014). Self-reported speech problems in adolescents and young adults with 22q11.2 Deletion Syndrome: A cross-sectional cohort study. Archives of Plastic Surgery. 41: 472-479.

Widdershoven, J.CC., Beemer, F.A., Kon, M., Dejonckere. & Van Der Molen, M. (2008). Possible mechanisms and gene involvement in speech problems in the 22q11.2 deletion syndrome. Journal of Plastic, Reconstructive and Aesthetic Surgery. 61, 1016-1023.