# SPEECH AND 22Q

AKSHAT SHAH, MAEVE MORRISON

SPEECH LANGUAGE THERAPISTS

CLEFT LIP AND PALATE TEAM, MIDDLEMORE HOSPITAL

## WHY SPEECH LANGUAGE THERAPISTS?

- Speech Language Therapists look at both feeding and communication
- Speech Language Therapist often first to suspect 22q in absence of cardiac anomaly
- Appropriate intervention at appropriate time
- Refer to paediatrician
- Refer to cleft team to consider speech surgery

## FEEDING AND COMMUNICATION

- Eating and drinking issues
- Impairment of verbal communication in 22q
- Velopharyngeal Impairment (VPI)
- Speech
- Language
- Pragmatics

## FEEDING IN 22Q

- Palate & pharyngeal anomalies (VPI)
- Cardiac anomaly
- Gastrointestinal (reflux, constipation)
- Airway anomalies
- Swallowing disorder

#### FEEDING ISSUES

- Sucking problem
- Nasal emission of food/fluid
- Swallowing disorder due to incoordination of suck- swallow-breathing pattern
- May require tube feeding for weeks/years
- Need specialised feeding to avoid aspiration and provide nutrition
- Fussy about textures and/or tastes

## MANAGEMENT OF FEEDING

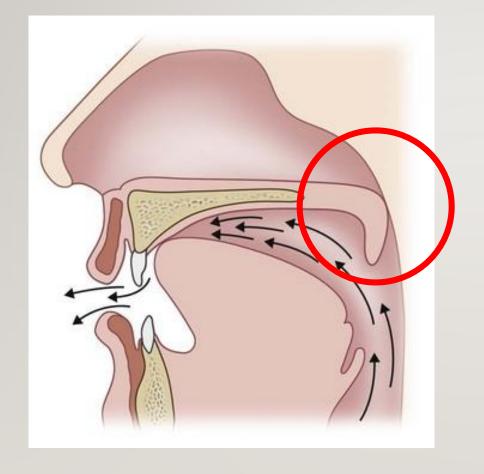
- Following assessment of swallow the SLT may provide/advise:
  - Specialised feeding bottles
  - Upright positioning
  - Thickened fluids may be easier than fluids
  - Sticky textures often more problematic

## COMMUNICATION

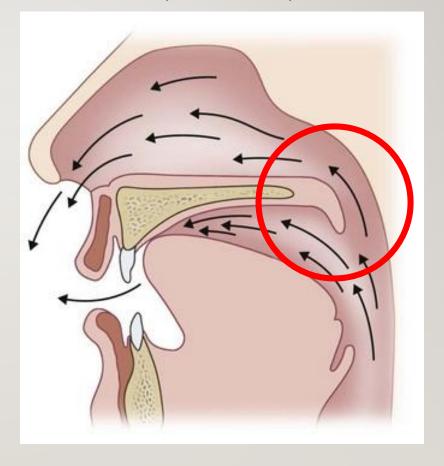
- Delayed onset of language
- Velopharyngeal Impairment
- Apraxia of speech

- Language delay
- Hearing impairment
- Social skills

### Airflow during typical speech



## Airflow during atypical speech (nasal tone)



## VELOPHARYNGEAL IMPAIRMENT (VPI)

- Hypernasal resonance
- Nasal emission on pressure sounds
- Cleft type speech substitutions
  - Glottal articulation (glottal stop for plosives /p b t d k g/)
  - Pharyngeal fricative for fricatives /s sh z f/
  - Backing pattern
  - Weakened plosives
  - Nasalised plosives/fricatives
- Food or fluid leaking out nose

## VPI IN 22Q

- As a result of :
  - Cleft palate
  - Submucous cleft palate
  - Large VP gap
  - Poor VP function

- Surgery for VPI
  - Cleft palate or submucous cleft palate repair
  - Pharyngoplasty
  - Pharyngeal flap
  - Pharyngeal implant
- Often delayed response to speech surgery

## SPEECH SURGERY IN 22Q

- Should reduce hypernasality
- Enable child to create oral pressure for speech sounds

#### Surgery is considered when:

- VPI is confirmed by videofluoroscopy or nasendoscopy
- Significant communication impairment
- Benefits of surgery outweigh the risks

#### BEHAVIOUR AND PRAGMATICS

- May have problems with anxiety
- Can be shy and withdrawn
- Can be impulsive
- May be oversensitive to failure
- May not always understand subtleties in language.
  Take things literally
- Can be obsessive /compulsive
- Can go on to have issues in teenage years and beyond

### MANAGEMENT OF SPEECH

- Early intervention for speech and language
- Working on placement of speech sounds
- Front sounds before back
- Word final might be easier than word initial
- Use gentle lengthened production of sound

## **WORKING WITH THE SLTS**

- Cleft Team SLTs and Community-based SLTS
- Therapy should be:
  - Individual
  - Specific
  - Regular with practise at home
  - Repetitive
  - Fun
  - May need signing

#### REFERENCES

Carneol, S.O., Marks, S.M., Weik, L.A. (1999). The speech-language pathologist: Key role in the diagnosis of velocardiofacial syndrome. *American Journal of Speech-Language Pathology, 8*(1), 23-32.

Cutler-Landsman, D. (2012). Educating children with velo-cardio-facial syndrome (2<sup>nd</sup> ed.). San Diego, USA: Plural Publishing.

Golding-Kushner, K.J. & Shprintzen, R.J. (2008). Velo-cardio-facial syndrome: Volume 1, San Diego, USA: Plural Publishing.

Golding-Kushner, K.J. & Shprintzen, R.J. (2011). *Velo-cardio-facial syndrome* treatment of communication disorder: Volume 2, San Diego, USA: Plural Publishing.

Scherer, N.J., D'Antonio L.L, & Kalbffleisch, J.H. (1999). Early speech and language development in children with velocardiofacial syndrome. *American Journal of Medical Genetics*, 88, 714-723.

Solot, C. B., Knightly, C., Handler, S. D., Gerdes, M., McDonalald-Mcginn, D. M., Moss, E....Larossa, D. (2000). Communication disorders in the 22q11.2 microdeletion syndrome. *Journal of Communication Disorders*, 33(3), 187- 204. doi:10.1016/S0021-9924(00)00018-6.