# Bridging the Gap: Sleep Disordered Breathing in Myelomeningocoele and ACII Malformation

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# Journey

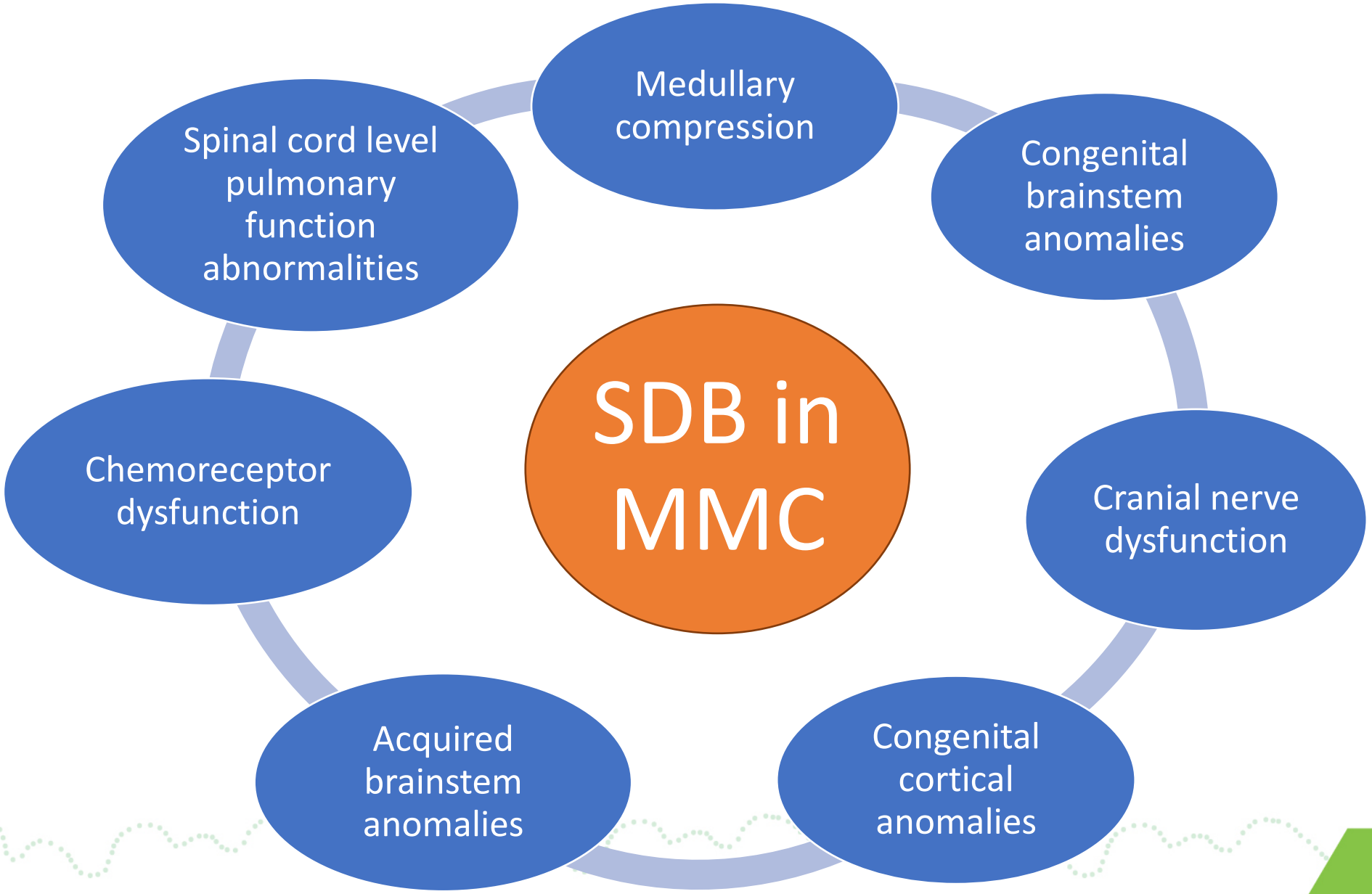
- Sleep disordered breathing and myelomeningocoele
- Audit
- Key Findings
- Interventions
- Future

# Pathogenesis

- Prevalence
  - 40 – 85%

- Presents…
- OSA
- CSA
- Mixed

- Asymptomatic disease a concern



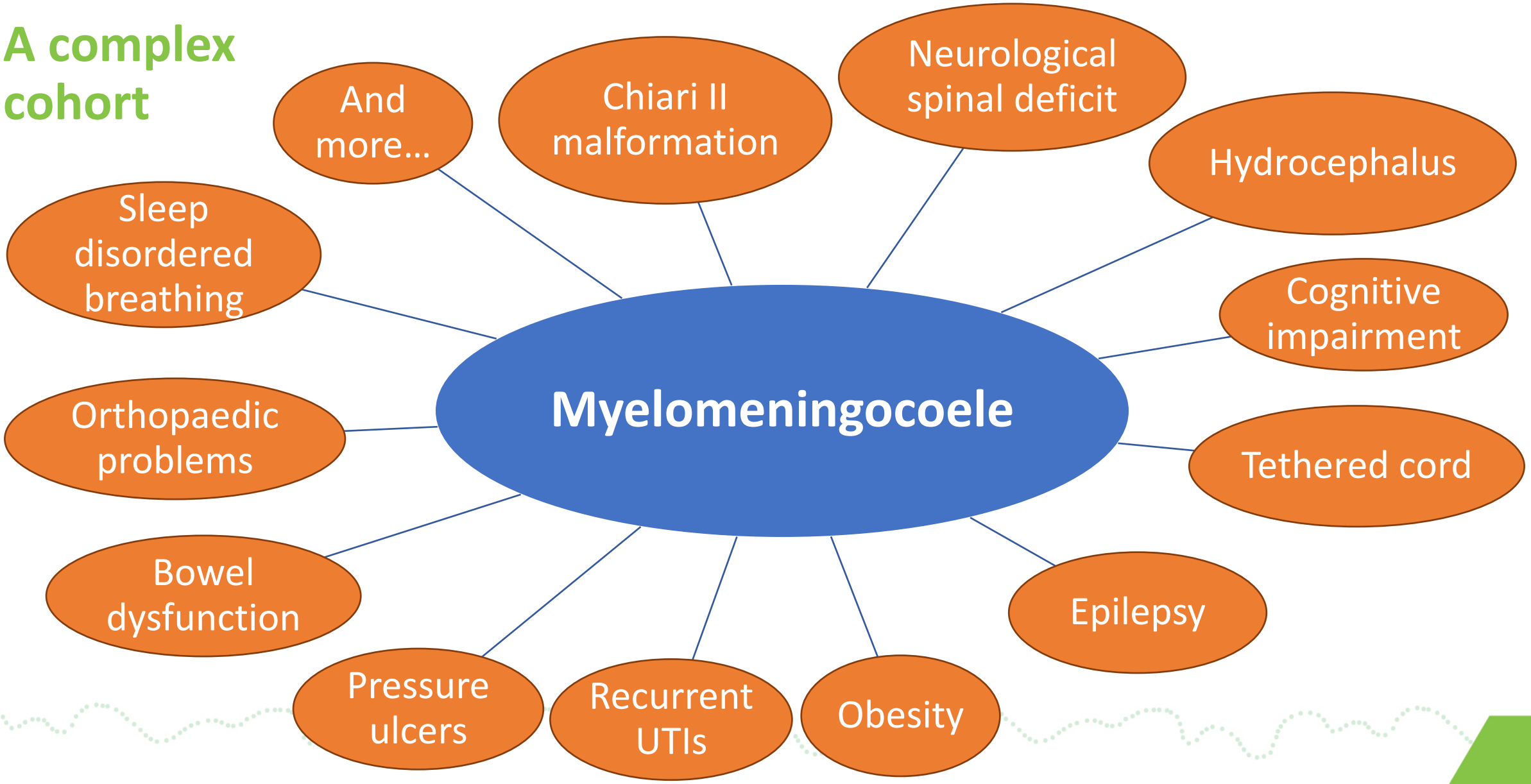
# Why does this matter?

- **Morbidity**
  - Contributory or causative?
- **Screening challenges**
- **Long term outcomes**
  - Paucity of data
  - ? Association with early mortality in un-diagnosed SDB

## Clinical consequences of SDB

- Neurocognitive and developmental
- Growth and metabolic dysfunction
- Cardiopulmonary and autonomic dysregulation
- Quality of life
- Survival impacts

# A complex cohort



# CHQ guidance recommendations (2023)
# SDB in children 0 – 3 years

*1) Overnight oximetry and urgent sleep study referral for infants with cervical and thoracic level lesion or symptoms significant for Arnold Chiari Malformation.*

*2) Consider continuous oxygen saturations overnight in the nursery before discharge as a screening tool and referral to respiratory team as required. This is a reasonable screening tool before referral for a formal sleep study.*

*3) A baseline sleep study is desirable in all children with Arnold Chiari malformation and those with thoracic lesions and above during first four weeks.*

## Audit Aims

1. Review clinician adherence with current guidelines.
    1. If present - address barriers to clinician adherence
    2. Implement changes
    3. Review after a period of 3 years

2. Assess relevance of the current local guideline with assessment of data
    1. Determine incidence of SDB if possible
    2. Impacts of referral timing, investigation and management

## Methods

- Retrospective chart audit at QCH

- Inclusions
    - Attending spinal disabilities clinic at QCH
    - Born between January 2018 and June 2023
    - MMC of thoracic or cervical origin OR Presence of AC2

- Key audited data: referral dates to the sleep clinic, indications given for PSG *, attendance at sleep appointments, and PSG results

*Polysomnography (PSG) / Sleep study

# Results: Referral Practices and Adherence

**26 children**

Eligible for audit

**50%**

Eligible cases were referred

**357 Days**

Average age at referral to sleep clinic

**477 Days**

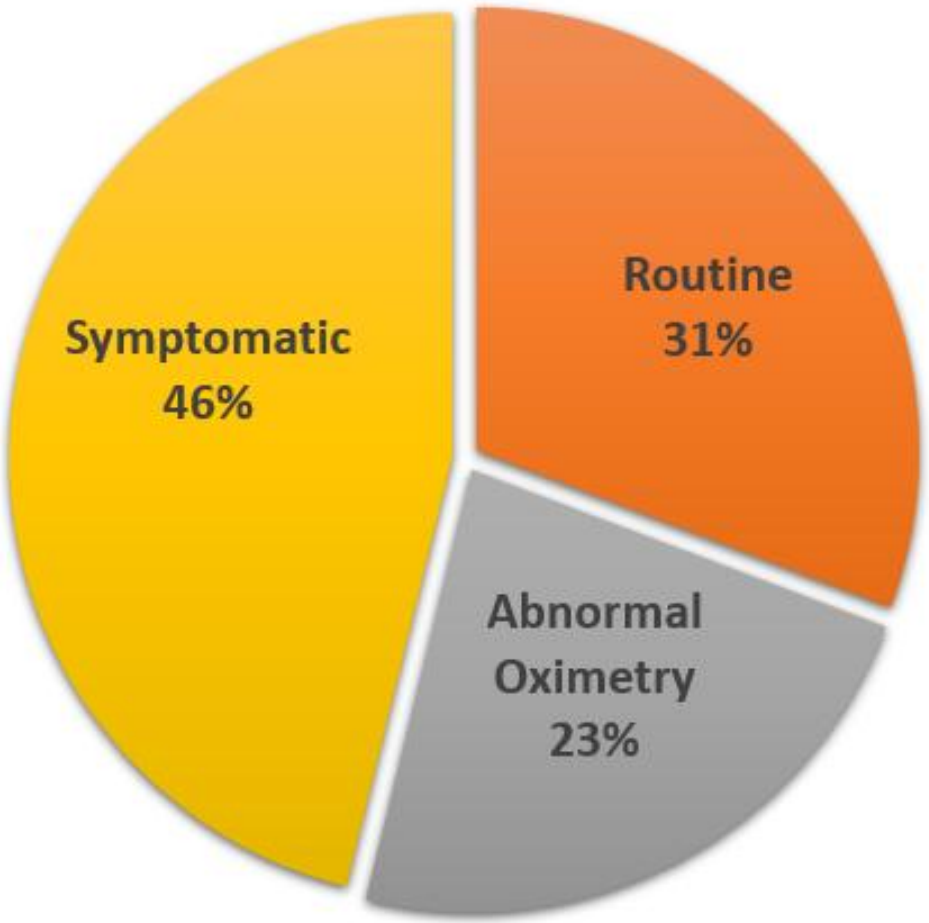
Average age at first PSG

**211 Days**

Average time between referral and PSG

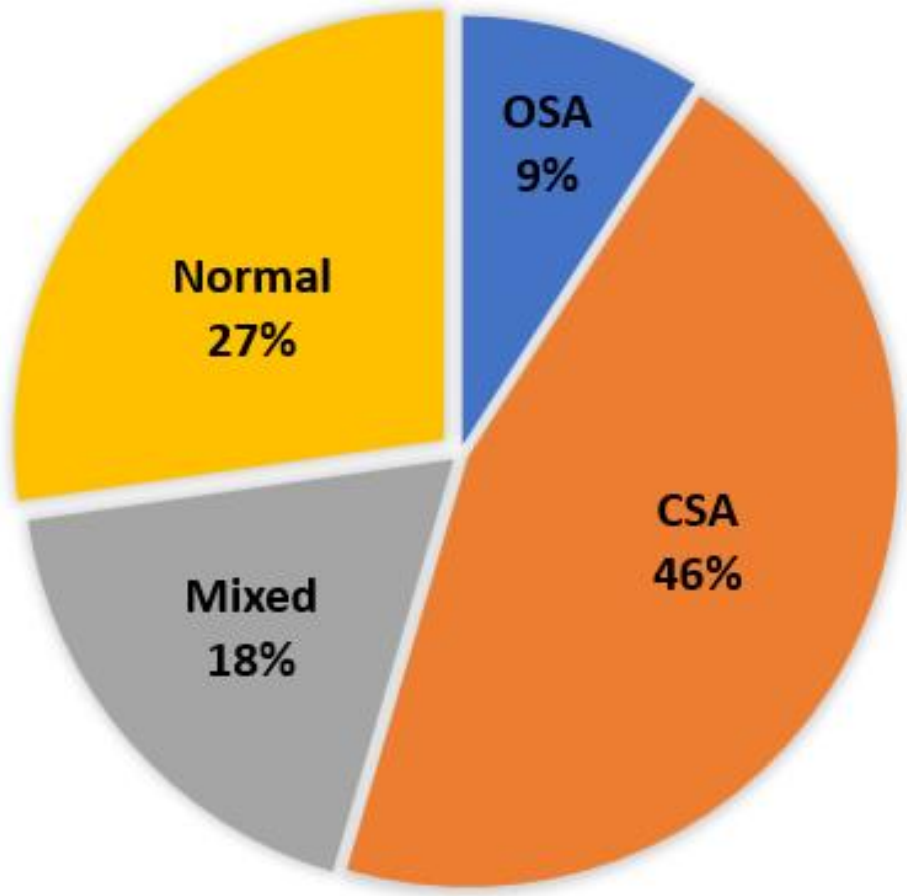
* Oximetry data

# Results: Referral Indications




- Symptomatic n = 6
  - Eg; snoring, arousals, apnoeas, daytime somnolence…
- Routine n = 4
- Abnormal oximetry n = 3

# Results: PSG findings




Children had PSG n = 11 / 13 referred
- Awaiting PSG at time of audit = 1
- PSG not performed at clinician discretion = 1

Demonstrated pathology n = 8 (72%)
- OSA = 1
- CSA = 5
- Mixed = 2

Required treatment n = 6 (54%)
- *Adenotonsillectomy recommended = 1*
- *Supplemental O2 = 4*
- *CPAP = 1*

# Reflections

- Data reflects significant deficiencies in screening practices

- Complex cohort
  - Competing clinical priorities
  - Time restraints

- Current referral practices are reactive rather than proactive
  - SDB may be undetected until symptoms appear

- Referral delays – combination of …
  - Clinician awareness gaps
  - Workforce and staffing challenges in public system
  - Limited appointment and sleep lab availability
  - Inefficiencies in referral and triage practice

# Proposed interventions and Future Directions

## Raise Awareness

- Elevate awareness of existing guidelines to key stakeholders

## 1) Automatic approved referrals

- Endorsement from respiratory medicine service regarding referrals
- Case made for other high-risk populations (Down syndrome and Prader Wili)

## 2) New Babies Checklist

- Ensure all routine referrals performed as early as possible
- Helmed by spinal disabilities CN

## 3) Improvements in EMR

- Improvements targeted in documentation between non iEMR sites / Nurseries
- Improved documentation of neonatal oximetry as per guideline: aim to improve triage process

# Limitations

- **Retrospective research**
  - Risk of inaccuracies

- **Referral** indications for PSG demonstrated **selection bias**
  - Children who were referred were more likely to be **symptomatic**, skewing the findings

- **Limited sample size** (**n=26**)
  - Restricts the ability to draw **statistically significant** conclusions

- **Only 50% of eligible children underwent PSG**
  - Prevalence of SDB in our cohort is unknown

# Summary

- SDB may be an underappreciated complication of MMC
- Screening processes challenging
- In this audit
  - Only 50% of children were referred for PSG screening
  - Delays in obtaining PSG also observed
  - Majority of those tested had abnormal findings; reinforcing need for proactive screening
- Changes; including structured checklists and improvements to documentation are being implemented
- A guideline is only worthwhile if it is followed
- Prompt reflection at own centers re: screening practices for SDB

# With thanks…

- Entire QPRS Spinal Disabilities team including…
  - Dr Lisa Copeland
  - Dr Owen Gilles
  - CN Jennifer Miller

- Key References

  - Gunnett M, Rocque BG, Nourani A, Beltran-Ale G. Impact of Spina Bifida on Sleep Quality: Current Insights. Nat Sci Sleep. 2023 Nov 24;15:967-978. doi: 10.2147/NSS.S401269. PMID: 38034043; PMCID: PMC10685378.
  - Liptak GS, Robinson LM, Davidson PW, Dziorny A, Lavalley R, Flaherty MG and D. NP. (2016). "Life course health and healthcare utilization among adults with spina bifida." Dev Med Child Neurol. **58**(7): 724-720.
  - Lazzareschi, I., et al., *Sleep-disordered breathing in patients with Chiari Malformation type II: a case-control study and review of the literature.* J Clin Sleep Med, 2022. **18**(9): p. 2143-2154.
  - Macaldowie A, H.L., *Neural tube defects in Australia: prevalence before mandatory folic acid fortification* M.a.M.U. Communications, Editor. 2011, Australian Institute of Health and Welfare: Canberra.
  - QPRS, D., *Staying Healthy: Infants and Children with Spina Bifida*, QPRS, Editor. 2023: Brisbane, Queensland, Australia