# WHY ARE PIERRE ROBIN SEQUENCE RATES HIGH IN SOUTH WALES?

HELEN EXTENCE

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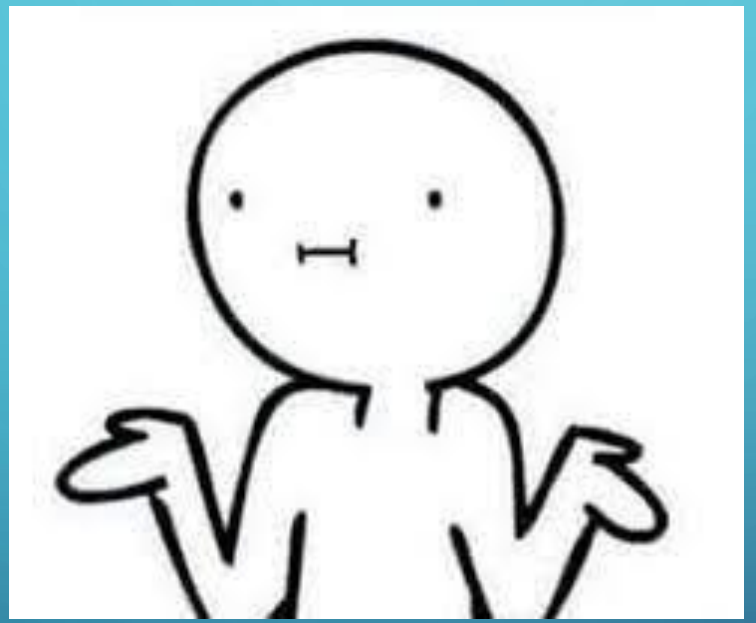
Wefus a Thaflod Hollt
The Welsh Centre for
Cleft Lip & Palate

GIG CYMRU NHS WALES
Bwrdd Iechyd Prifysgol Bae Abertawe
Swansea Bay University Health Board

I don't know



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- Positive outlier for the proportion of children with Cleft palate only who were reported to have PRS for births 2020-2023 (46 children with Cleft palate)
- Status is known for each child

| South Wales | National rate |
|---|---|
| 48% 22 children PRS, 24 absent | 21% |

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| Cleft Service | Children with cleft palate | Reported RS status | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|
| | | RS status reported | | RS present | | RS absent | | RS status unknown | | RS assumed to be asbsent | |
| | N | n | (%) | n | (%) | n | (%) | n | (%) | n | (%) |
| Newcastle | 51 | 51 | 100.0% | 11 | 21.6% | 40 | 78.4% | 0 | 0.0% | 40 | 78.4% |
| Leeds | 72 | 71 | 98.6% | 9 | 12.5% | 62 | 86.1% | 1 | 1.4% | 63 | 87.5% |
| Liverpool | 66 | 48 | 72.7% | 5 | 7.6% | 43 | 65.2% | 18 | 27.3% | 61 | 92.4% |
| Manchester | 67 | 65 | 97.0% | 14 | 20.9% | 51 | 76.1% | 2 | 3.0% | 53 | 79.1% |
| Trent | 94 | 92 | 97.9% | 17 | 18.1% | 75 | 79.8% | 2 | 2.1% | 77 | 81.9% |
| West Midlands | 98 | 93 | 94.9% | 29 | 29.6% | 64 | 65.3% | 5 | 5.1% | 69 | 70.4% |
| Cleft Net East | 68 | 8 | 11.8% | 4 | 5.9% | 4 | 5.9% | 60 | 88.2% | 64 | 94.1% |
| North Thames | 138 | 128 | 92.8% | 36 | 26.1% | 92 | 66.7% | 10 | 7.2% | 102 | 73.9% |
| Spires | 96 | 96 | 100.0% | 10 | 10.4% | 86 | 89.6% | 0 | 0.0% | 86 | 89.6% |
| South Wales | 46 | 46 | 100.0% | 22 | 47.8% | 24 | 52.2% | 0 | 0.0% | 24 | 52.2% |
| South West | 43 | 37 | 86.0% | 8 | 18.6% | 29 | 67.4% | 6 | 14.0% | 35 | 81.4% |
| Evelina London | 128 | 115 | 89.8% | 27 | 21.1% | 88 | 68.8% | 13 | 10.2% | 101 | 78.9% |
| Northern Ireland | 40 | 35 | 87.5% | 12 | 30.0% | 23 | 57.5% | 5 | 12.5% | 28 | 70.0% |
| Scotland | 25 | 25 | 100.0% | 9 | 36.0% | 16 | 64.0% | 0 | 0.0% | 16 | 64.0% |
| **Total** | **1,032** | **910** | **88.2%** | **213** | **20.6%** | **697** | **67.5%** | **122** | **11.8%** | **819** | **79.4%** |

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# AS A CENTRE…

Q1. Are we better at identifying PRS ?

Q2. Are we better at reporting PRS?

Q3. Are we over reporting PRS ?

Q4. Do we have higher rates of PRS ?

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# Q1. BETTER AT IDENTIFYING?

- Clear pathway for these babies
- Managed in partnership with the Paediatric Respiratory Service Children's Hospital for Wales (CHfW), Cardiff since 2019

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## Assessment and Referral

**Referral**
Refer to cleft team: 01792 703810 [service will make contact within 24 hours]
Consider transfer baby to neonatal unit
NG feeds and Nil by mouth
Oximetry and monitoring

- Assessment by Cleft and Neonatal teams
- Features consistent with Pierre Robin Sequence or other micrognathia syndromes
- Discuss with Paediatric Respiratory Service @ The Children's Hospital for Wales

| Respiratory distress | Some respiratory concerns | No respiratory concerns |
|---|---|---|
| Oximetry and monitoring<br>Insert nasopharyngeal airway | NG feeds<br>Nil by mouth<br>Oximetry and monitoring<br>Optimise position (side) | Overnight oximetry study<br>Discuss with Paediatric Respiratory service @ Children's Hospital for Wales |
| NG feeds<br>Nil by mouth | Referral and transfer to Paediatric Respiratory Service @ Children's Hospital for Wales for 10 day admission | Abnormal oximetry study |
| Referral and transfer to Paediatric Respiratory Service @ Children's Hospital for Wales for 10 day admission | | NG feeds<br>Nil by mouth<br>Oximetry and monitoring<br>Optimise position (side) |
| | | Referral and transfer to Paediatric Respiratory Service @ Children's Hospital for Wales for 10 day admission |
| | | Normal oximetry study (consider repeating Day 5) |
| | | Feeding assessment by Cleft Nurse<br>Training and provision of feeding equipment |
| | | Equipment Provision and training [Home Oximetry, BLS, car seat challenge] |
| | | Audiology assessment<br>Ophthalmology assessment<br>Echocardiography<br>Check antenatal ultrasound for renal abnormalities |
| | | Discuss with Paediatric Respiratory Service @ Children's Hospital for Wales prior to discharge |
| | | Sleep study @ Children's Hospital for Wales within 2 weeks |

## Pre-cleft repair. Respiratory service management and timeline.

### Assessment at birth

Sleep polygraphy without NPA for assessment of obstruction
Insertion of Vygon Nasopharyngeal airway [stick to a single nostril]
Sleep study with NPA in-situ for assessment of NPA positioning

Nil by mouth and NG feeds
Equipment Provision and training [NPA management, Home Oximetry, suction, BLS, car seat challenge]
Feeding assessment by SLT. Training and provision of feeding equipment [liaison with local services]
Audiology assessment, Ophthalmology assessment, Echocardiography, Renal assessment
Airway management information leaflet for parents
Respiratory / Cleft MDT

### Assessment at 3 months

Reassess size of NPA
Sleep polygraphy with newly sized NPA in-situ
Nil by mouth and NG feeds
Feeding assessment by SLT. Potential to start remove then replace NPA for feeds during the day

### Assessment at 6 months

Reassess size of NPA
Sleep polygraphy overnight with NPA in-situ, then extend into daytime with NPA removed
If polygraphy acceptable through daytime naps, remove NPA during the day and reinsert each night
Feeding assessment by SLT. Increase oral feeding as possible

### Assessment at 9 months

Sleep polygraphy overnight with NPA removed
If polygraphy acceptable, remove NPA both day and night
Parents keep NPA kit in case of problems with intercurrent infection
Parents continue nightime oximetry monitoring until after cleft palate repair

### Assessment at 14 months [post cleft repair]

Sleep oximetry post cleft palate repair
If oximetry successful, remove home oximetry monitoring
Discharge from paediatric respiratory and sleep service

# REFLECTIONS OF OUR CNS

- "Prior to 2019, mild to moderate PRS were managed by the cleft team through NG and positioning but felt was not as well documented and **possibly under reported"**
- "Prior to this pathway we would only refer the babies who were showing the above features and signs of respiratory distress or obstruction but now most babies are referred, we have found that occasionally babies who look like they are managing their airway are actually having obstructive events, some requiring an NPA"

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# Q2. BETTER AT REPORTING PRS?

- Good system in place
- PRS diagnosis- written on the baby clinic letters, operation notes, baby registration card so easy to enter the data
- One member of the team is responsible for inputting onto CRANE

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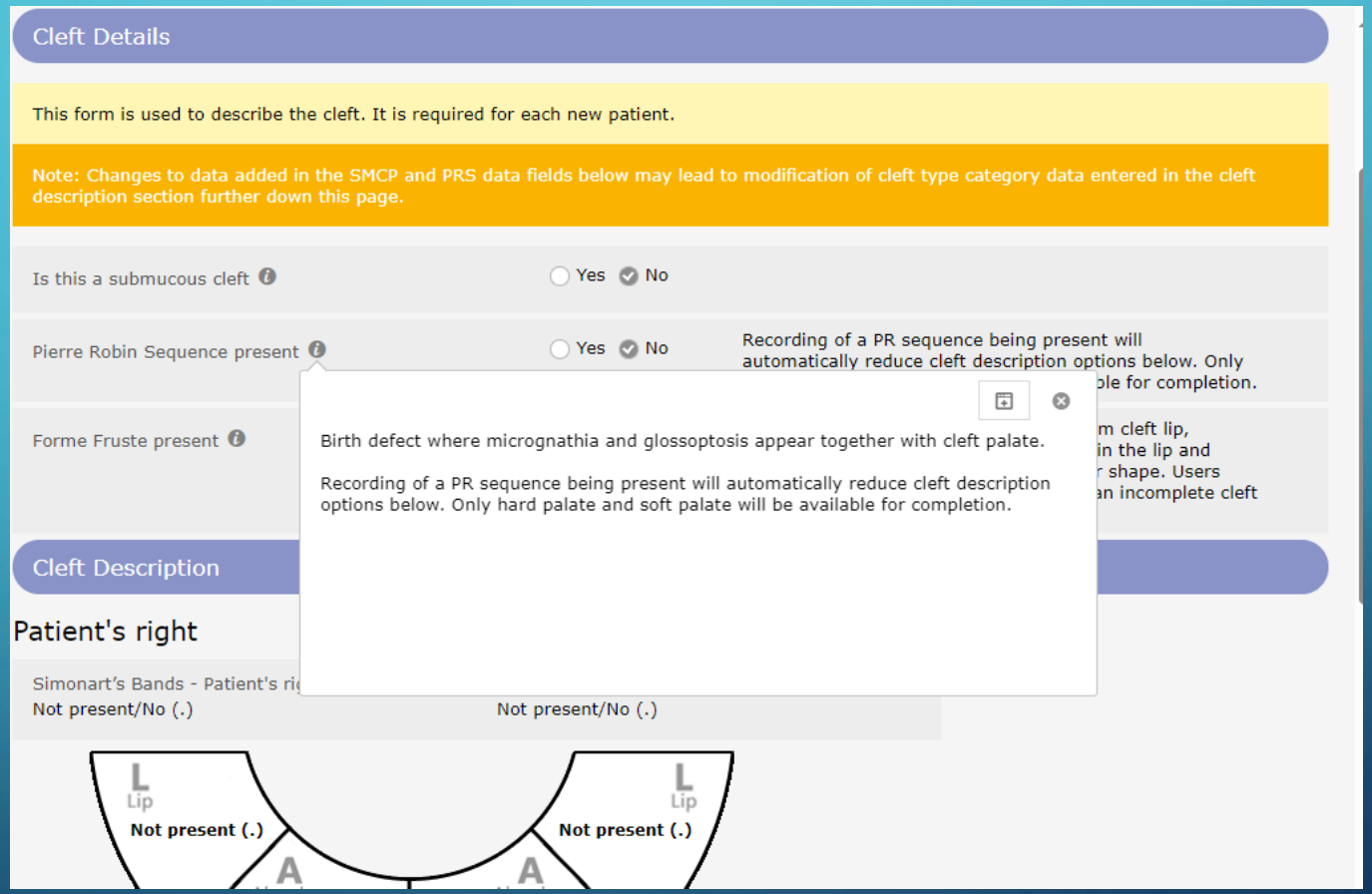
# Q3. OVER REPORTING PRS ?



| GIG CYMRU NHS WALES — Bwrdd Iechyd Prifysgol Bae Abertawe / Swansea Bay University Health Board | Hospital No: | CRANE Identifier: | Surgeon: |
|---|---|---|---|

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## New Baby Registration Form

| | | | |
|---|---|---|---|
| Surname: | | Date and time of birth: | |
| Surname at birth: | | Sex: | |
| First names: | | E.D.D: | |
| Address including postcode:<br><br>Home Phone Number: | | | |
| G.P Name and Address: | | | |
| Community Midwife: | | | |
| Health Visitor: contact number & email address | | | |
| Social Worker/ other professionals: | | | |
| Mothers Name, Mobile & email address:<br><br>(Address if different) | | Fathers Name, Mobile & email address:<br><br>(Address if different) | |
| Next of Kin: | | | |

Correspondence letters to be sent to both parents if addresses differ? Yes/No

| Ethnic group: (2001 Census classification) | | | |
|---|---|---|---|
| 1. Asian/Asian British- Indian | | 9. Other Black | |
| 2. Asian/Asian British- Pakistani | | 10. Chinese | |
| 3. Asian/Asian British- Bangladeshi | | 11. White- Irish | |
| 4. Other Asian | | 12. Mixed- white& Black Caribbean | |
| 5. White- British | | 13. Mixed- White & Black African | |
| 6. Other Ethnic group | | 14. Mixed- White & Asian | |
| 7. Black/Black British- Caribbean | | 15. Other Mixed | |
| 8. Black/Black British- African | | 16. White- other | |
| Any other essential information (eg. Interpreter required) | | Yes | No |

| | | | |
|---|---|---|---|
| Cleft Description: | | | |
| Diagnosed by whom: | | | |
| Date and time of Referral: | | Referring Professional & Hospital: | |
| Paediatrician: | | | |
| Date and time first seen: | | Geneticist? Y/N<br>Name of Geneticist | |
| Prenatal diagnosis of cleft? Y/N | | Home Birth? Y/N | |
| Delivery: (natural, cesarean, forceps, ventouse, breach) | | Reason if not natural delivery: | |

1.Single ☐ 2. Identical twin ☐ 3. Non-identical Twin ☐ 4. More ☐

If Twins, Other twin affected? Yes/No

| | | |
|---|---|---|
| Gestation: /40 | Weight (kg): | Head Circumference (cm): |

Timing of Diagnosis:

| | | | |
|---|---|---|---|
| Antenatal | | Pierre Robin Sequence?<br>PRS (micrognathia + Glossoptosis + cleft palate | |
| At birth (within 24 hours of birth) | | | |
| Within 72 hours | | PRS severity: | |
| Within 1 week | | Sub mucous cleft: | |
| Within 1 month | | Other Syndrome: | |
| Within 6 months | | | |
| Later than 6 months | | | |
| Family Cleft History? Y/N | | Describe- (e.g. mother cleft lip) | |
| Family Medical History? Y/N | | Describe: | |

Older Siblings

| | 1 | 2 | 3 | 4 | 5 | 6 |
|---|---|---|---|---|---|---|
| Child Name | | | | | | |
| DOB | | | | | | |
| Sex | | | | | | |

# Q4. DO WE HAVE HIGHER RATES OF PRS ?

- “Seem to have more PRS Palate babies born and less isolated straightforward cleft palate babies”
- Despite a very small birth year of 26 babies last year, out of our 10 palates, 8 had PRS
- Our lead in genetics, is not aware that we have a higher burden of genetic conditions causing PRS

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- Tom Jovik, Specialist registrar has been looking at the geographical epidemiology of CLP in South Wales
- Using the CRANE database 2013- 2021 and the Office of National statistics database
- PRS had an incidence of 2.2 per 10,000 live births in South Wales
- Our incidence of PRS is almost double the globally cited incidence of this anomaly

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# I STILL DON'T KNOW… BUT…..

- Need to go back to 2020 data and verify that all cases meet the definition by CRANE
- With our pathway in place, we have good identification of PRS, good admin, reporting and inputting systems in place
- Warrants further discussion with our respiratory colleagues, paediatricians and genetic colleagues
- Look at genetic data and deprivation scores

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# THANK YOU FOR LISTENING

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