# Clinic For Children With Rare Diseases

## A Bevan Exemplar Project


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# Background

- Rare disease is defined as any condition with an incidence of less than 1:2000 in the population
- Up to 7000 distinct conditions recognized
- Estimated more than 200 new conditions identified each year
- Approximately 150.000 people in Wales affected
- 1:17 of us will have a rare disease at some time in our life
- **Rare disease is individually rare but collectively common!**

# Background

- Often complex, multisystem, life limiting, multiple care needs
- Lack of understanding by health care professionals
- Health care inequalities
  - Lack of provisions
  - Suboptimal emergency management
  - Lack of appropriate surveillance
  - Missing out on new and emerging treatments
- National strategy: Wales Rare Disease Action Plan 2022 -2026

# Aims of the project

- Develop a multidisciplinary clinic for children with rare disease
- Identify children with complex rare disease in the catchment area of a Paediatric unit in Wales
- Enable patients and their carers to ‘understand their disease’
- Improve coordination of care
- Improve access to specialist care, treatments and drugs

# Approach

- Princess of Wales Hospital Bridgend serves as pilot
- All patients under current paediatric care reviewed – patients fulfilling key criteria of disease incidence less than 1:2000 identified
- CARIS (Congenital Anomaly Register and Information Service) data cross checked
- Notifications through local team
- **137 children with a rare disease and complex needs identified**

# Outline of clinic

- Multidisciplinary clinic run in partnership with parents and carers
- Pre clinic extensive search of databases and medical literature
- 2-hour appointment
- Surveillance needs and time intervals defined, documented and actioned
- Emergency measures defined and documented

# Outline of clinic

- Information patients and carers bring to the appointment i.e. via social media discussed
- Patients signposted to patient support groups
- Referral and research centres identified and approached
- Transition to adult services initiated if appropriate
- Follow up communication via appointment, telephone or home visit as per families' choice

# Impact measures

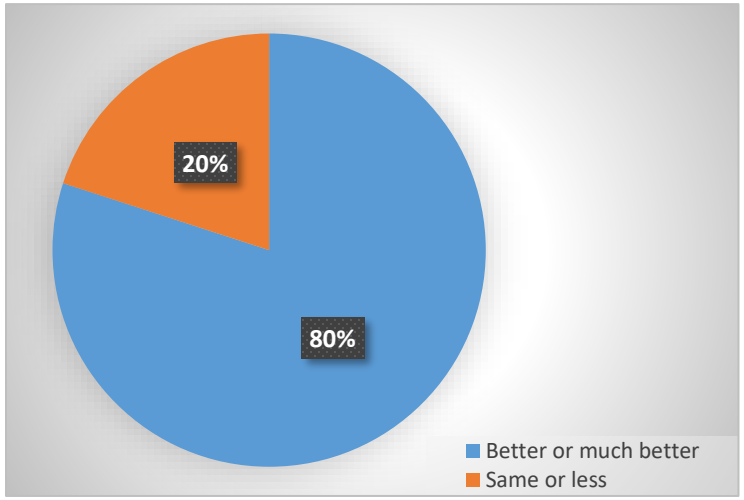
- Patient Reported Outcome Measures (PROMs)
  - Assess quality of care delivered from the patient's perspective (ongoing)
- Patient Reported Experience Measures (PREMs)
  - Capturing patients' experience and perception (ongoing)
- Family Reported Outcome Measures (FROMs)
  - Measuring the impact on the quality of life of an adult family member (planned)

# Results

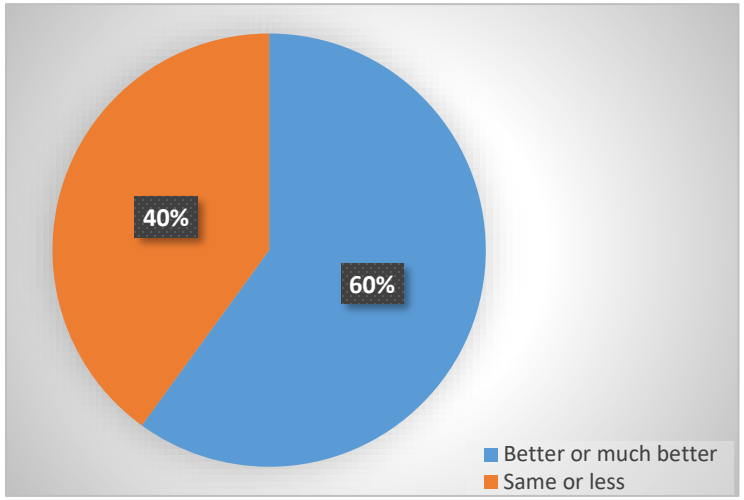
- 10 patients seen
- PROMs: 5 patients
- PREMs: 4 patients

# PROMs (5 patients and parents)
# Patient Enablement Instrument

**Able to understand your child's illness**




**Able to cope with life**
**Able to cope with your child's illness**
**Able to keep your child healthy**
**Confident about your child's health**
**Able to help your child**



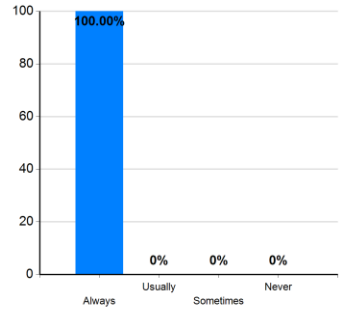
# PREMs (4 patients and parents)

## Did you feel that you were listened to?

**Question 1:** Did you feel that you were listened to?

| Available Answers | Responses | Score (%) |
|---|---|---|
| Always | 4 | 100.00% |
| Usually | 0 | 0.00% |
| Sometimes | 0 | 0.00% |
| Never | 0 | 0.00% |
| **Total** | **4** | **100%** |

Create new action




## From the time you realised you needed this service, was the time you waited?

**Question 3:** From the time you realised you needed to use this service, was the time you waited:

| Available Answers | Responses | Score (%) |
|---|---|---|
| Shorter than expected | 3 | 75.00% |
| About right | 1 | 25.00% |
| A bit too long | 0 | 0.00% |
| Much too long | 0 | 0.00% |
| Not applicable | 0 | 0.00% |
| **Total** | **4** | **100%** |

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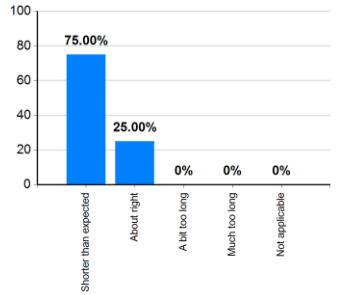


## Did you feel well cared for?

**Question 2:** Did you feel well cared for?

| Available Answers | Responses | Score (%) |
|---|---|---|
| Always | 4 | 100.00% |
| Usually | 0 | 0.00% |
| Sometimes | 0 | 0.00% |
| Never | 0 | 0.00% |
| **Total** | **4** | **100%** |

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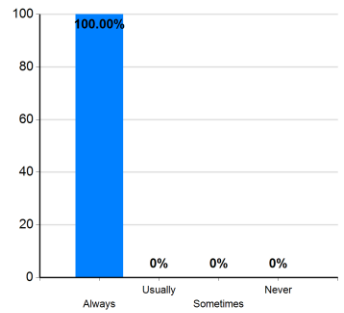


## If you asked for assistance, did you get it when you needed it?

**Question 4:** If you asked for assistance, did you get it when you needed it?

| Available Answers | Responses | Score (%) |
|---|---|---|
| Always | 4 | 100.00% |
| Usually | 0 | 0.00% |
| Sometimes | 0 | 0.00% |
| Never | 0 | 0.00% |
| Not applicable | 0 | 0.00% |
| **Total** | **4** | **100%** |

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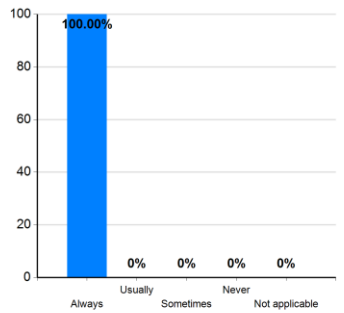


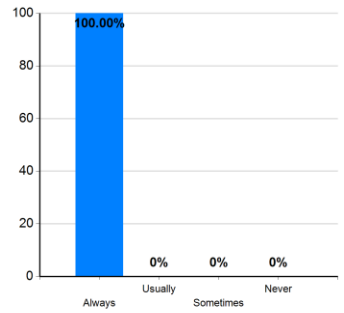
# PREMs (4 patients and parents)

## Did you feel you understood what was happening?

**Question 5:** Did you feel you understood what was happening?

| Available Answers | Responses | Score (%) |
|---|---|---|
| Always | 4 | 100.00% |
| Usually | 0 | 0.00% |
| Sometimes | 0 | 0.00% |
| Never | 0 | 0.00% |
| **Total** | **4** | **100%** |

Create new action

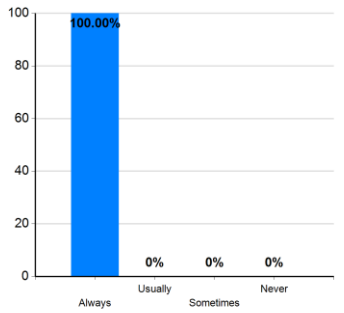



## Were you involved as much as you wanted to be in making decisions?

**Question 7:** Were you involved as much as you wanted to be in making decisions?

| Available Answers | Responses | Score (%) |
|---|---|---|
| Always | 4 | 100.00% |
| Usually | 0 | 0.00% |
| Sometimes | 0 | 0.00% |
| Never | 0 | 0.00% |
| **Total** | **4** | **100%** |

Create new action

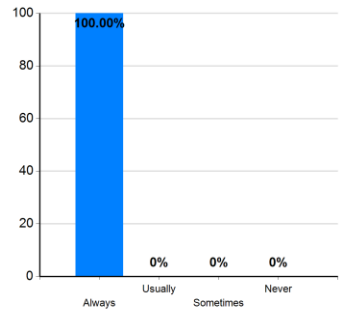



## Were things explained to you in a way you could understand?

**Question 6:** Were things explained to you in a way that you could understand?

| Available Answers | Responses | Score (%) |
|---|---|---|
| Always | 4 | 100.00% |
| Usually | 0 | 0.00% |
| Sometimes | 0 | 0.00% |
| Never | 0 | 0.00% |
| **Total** | **4** | **100%** |

Create new action

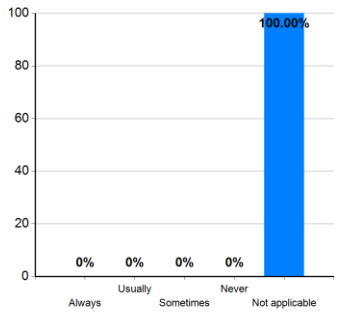



## Were you able to speak in Welsh to staff if needed?

**Question 8:** Were you able to speak in Welsh to staff if you needed to?

| Available Answers | Responses | Score (%) |
|---|---|---|
| Always | 0 | 0.00% |
| Usually | 0 | 0.00% |
| Sometimes | 0 | 0.00% |
| Never | 0 | 0.00% |
| Not applicable | 4 | 100.00% |
| **Total** | **4** | **100%** |

Create new action



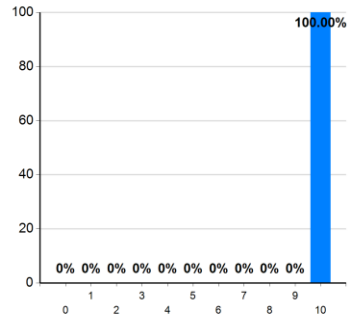
# PREMs (4 patients and parents)

**Using a scale of 0-10 where 0 is very bad and 10 is excellent, how would you rate your overall experience?**

**Question 9:** Using a scale of 0-10 where 0 is very bad and 10 is excellent, how would you rate your overall experience?

| Available Answers | Responses | Score (%) |
|---|---|---|
| 0 | 0 | 0.00% |
| 1 | 0 | 0.00% |
| 2 | 0 | 0.00% |
| 3 | 0 | 0.00% |
| 4 | 0 | 0.00% |
| 5 | 0 | 0.00% |
| 6 | 0 | 0.00% |
| 7 | 0 | 0.00% |
| 8 | 0 | 0.00% |
| 9 | 0 | 0.00% |
| 10 | 4 | 100.00% |
| **Total** | **4** | **100%** |

Create new action

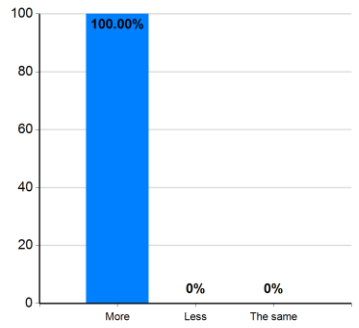



**Has the attendance in the Rare Disease Clinic made you feel more or less informed about your/your child's condition?**

**Question 10:** Has the attendance in the Rare Disease Clinic made you feel more or less informed about your/your child's condition?

| Available Answers | Responses | Score (%) |
|---|---|---|
| More | 4 | 100.00% |
| Less | 0 | 0.00% |
| The same | 0 | 0.00% |
| **Total** | **4** | **100%** |

Create new action



**Has the attendance in the Rare Disease Clinic made you feel more or less informed about your/your child's condition?**

- *I have had plenty of information on my daughter's condition and feel supported moving forward with whatever she may need*

- *We were provided with plenty of written information about our child's condition, along with links for further reading. A follow up appointment was arranged for 2 weeks after our initial appointment, in which our child's doctor was able to answer any questions we had about our child's condition*

- *I hadn't looked  in my child's condition since she was a baby. Things had changed in 16 years and her consultant showed us information we hadn't seen. He clearly looked into her condition thoroughly!*

- *We discussed my worries and fears for the future*

**Was there anything particularly good about your experience you would like to tell us about?**

- *I think the extra care and attention for children like my daughter is such an amazing service, support to make sure they are looked after in their life and us parents are supported along the way too*

- *Our child has been reluctant to attend clinic himself due to neurodevelopmental issues which are part of his condition. The clinical staff from the rare disease clinic involved in his care have been nothing but supportive regarding this, referring onwards to appropriate services and offering alternative provisions to accommodate his needs. This has made what has been a difficult time for us as a family much easier*

- *The consultant went above and beyond. He referred us to different departments which have been life changing*

- *I felt I was listened to and I appreciate the time spent with me*

# Would you recommend this service? Why/why not?

- *Yes, I think every family needs to feel heard, supported and gain knowledge to best understand and support their children*
- *Yes, we would recommend. It has been very convenient to have a 'one stop shop' for any issues arising from our son's condition, from which all relevant referrals have been made. Having a point of contact at rare disease clinic means we have been able to have any questions answered in a timely manner, and from the point of diagnosis we felt supported as a family*
- *Yes, because I feel all families with rare genetic disorders should receive the same care*
- *Yes, I would recommend this service as I feel a lot calmer now*

# Conclusion and next steps

- 10 patients seen so far
- Clinic set up in place and functional
- Feedback from patients, parents and carers very positive
- Expand, Spread and Adopt

# Thank you to:

- **Patients, parents and carers** for their participation and feedback
- **Local teams** for their support and encouragement
- **The Bevan Commission** for sponsorship and guidance
- **CARIS (Congenital Anomaly Register and Information Service)** for freely sharing data and expertise
- **WRDIN (Welsh Rare Disease Implementation Network)** for integration in the national strategy and help to deliver the plan within the Health Board

# Thank you!