

Improving care for patients with Gorlin Syndrome

Outcome & Research

STRENGTHS Research

WHY HAVE WE CHOSEN THIS CATEGORY OF PATIENTS

Gorlin syndrome is a **rare inherited disorder** characterized by **multiple basal cell carcinomas (BCC)** and **jaw keratocysts**. Other significant complications include **childhood medulloblastoma**, **ovarian and cardiac fibromas**, **ocular anomalies**, and **cleft lip and palate**.

One of the main defining features of Gorlin syndrome is the **early onset** of multiple BCCs, with a **median age of onset of 25 years**. Patients can have many hundreds of BCCs, leading to numerous **surgical and non-surgical therapies** and **disfigurement and scarring**. Many patients can **lose eyes, nose and ears** to their tumours. Often, patients are so badly affected that no treatment is possible to help with the lesions, leaving patients to manage with **ulcerated tumours** in **highly visible locations on the face**.

Many examples exist of patients **disengaging with health service providers** because of this and being lost to follow up resulting in **huge disfigurement** and **reduction of quality of life** and **social isolation** and occasionally **death** from the disease.

SWOT ANALYSIS

	Helpful to achieving the objective	Harmful to achieving the objective
Internal origin (attributes of the organization)	S Strengths	W Weaknesses
External origin (attributes of the environment)	O Opportunities	T Threats

WEAKNESSES Research

Across the country, patients are managed in various specialities in numerous different ways with no standardisation. This includes dermatology, plastic surgery, oncology, head and neck, oculoplastic, paediatric, and ear, nose and throat ENT services. Most dermatologists look after fewer than 10 patients.

This leads to differences in service access and access to treatments with wide variation in patient satisfaction across the country. Patients may be undertreated for their BCC or over treated with surgical procedures depending on the local experience and expertise of the specialists treating them. The patients are seen individually in single non-multidisciplinary clinics making it difficult for specialists to liaise when making treatment decisions. Furthermore, there are no established guidelines.

Patients are often discharged from clinics once they have had treatment, leading to late re-presentation with advanced BCC which can be very difficult and costly to treat. In some areas patients are discharged after treatment, they then have to go to GP for referral to radiology for annual screening. They are then dependant on the GP and radiologists to make a correct diagnosis.

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OPPORTUNITIES Research

In Manchester, after discussion with our patients and the **national Gorlin patient group**, we have set up the UK's **only multidisciplinary clinic** for Gorlin patients, with **dermatologists, geneticists, plastic surgeons, oncologist** and **specialist nurses** to address many of the above issues. Nowhere else in the UK offers this service. However, the specialist multidisciplinary clinic **serves only a fraction of the estimated 250** Gorlin patients in **Greater Manchester**.

The specialist multidisciplinary clinic has been **well received** by the patients that have been able to access it. The **Manchester Cancer Skin Pathway Board** aims to assess the two different models of care for Gorlin patients in Greater Manchester with a view to developing a case for **the formalisation and expansion** of the specialist multidisciplinary **clinic model**.

The Skin Pathway Board will assess its local Gorlin patients to see what **quality and experience of care** they are currently receiving and to **compare** those not in the dedicated clinic to those who have been seen in the clinic. If the clinic results in **better quality of care** and is perceived to be useful then we would **approach the commissioners** to expand the service to accommodate more local patients. We will also assess the need and demand for clinical **psychological care** in these patients and the specialist clinic.

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OPPORTUNITIES Research

We aim to:

- **Assess the quality of care** provided for Gorlin patients in Greater Manchester both within the multidisciplinary clinic and those not seen in this clinic **by validated questionnaire assessment** of patients.
- **Ask the patients for their views** on how the service for them should look and **what needs to improve.**
- Assess the need for **formal clinical psychological** input into these clinics
- Undertake analysis of the **cost effectiveness** of the specialist clinic model.
- Use the data obtained to **inform discussions** with Manchester Cancer, **commissioners** and **patients.**

Survey and Registry of Basal Cell Carcinoma Nevus Syndrome (BCCNS)
Gorlin Syndrome (GS) Individuals

This survey has been created to collect comprehensive data on individuals with BCCNS with the goal of helping the healthcare community better understand this condition and aim to further shape the service delivery for future care. The information gathered will be used to improve advocacy efforts and potentially accelerate the development of new treatments for individuals affected by Gorlin Syndrome. By participating in this registry and providing these valuable insights, you are making a significant and important contribution.

If you have already been registered on our database you will be asked a series of questions about your medical history and experience. If you are however new to our register you will be given the choice to leave your contact details for us to contact you to enable us to add you to our database. Please bear in mind the more information we can gain the better we can use this to understand, improve, (re)use your treatment and experience of services already provided. Please answer each question as completely and accurately as possible.

This questionnaire will take around 15 minutes to complete. Please fill in the relevant answers as best as you can by either ticking (✓) the selection or filling in the blank boxes. Should you have any queries about this questionnaire please contact louise.laurence@christie.nhs.uk. We thank you for your valued participation.

Q1. Can you provide the unique code that was sent with your paper questionnaire?

Yes
 No

If Yes, please enter the code here:

If No, please enter your telephone number so we can contact you:

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PERSONAL DETAILS

Q2. What is your age?

Under 18 18 to 24 25 to 34
 35 to 44 45 to 54 55+

Q3. What is your gender?

 

Q4. Where in the UK are you currently based?

South West
 South East
 East Anglia
 West Midlands
 East Midlands
 Yorkshire / Humber/Leeds
 North West
 North East
 Scotland
 Wales
 Northern Ireland

Q5. Over your lifetime have you resided in any other locations for a period of more than 2 years?

Location	Number of Years
Please list your country here:	

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PATIENT SATISFACTION

Please fill in / tick if relevant

Q25. On a scale of 1 to 10, how would you rate your current quality of life?

1	2	3	4	5	6	7	8	9	10
Poor							Neutral		Excellent

Q26. On a scale of 1-10 how would you rate the severity of your disease?

1	2	3	4	5	6	7	8	9	10
Not Severe							Severe		Very Severe

Q27. In your opinion how has your condition progressed within the last decade?

1	2	3	4	5	6	7	8	9	10
Greatly Improved				Stayed the Same					Greatly worsened

Q28. Have you used any health or support services for Gorlin Syndrome, this can be GP and/or specialist services?

Yes
 No

Q29. Can you give the approximate date you last used one of these services?

Q30. What if any, specialist services did you access? (You can tick more than one)

Dermatology / skin care
 Surgical procedure/s
 Photo dynamic therapy (PDT)
 Genetic counselling
 Dentist (Jaw cysts)
 GI (Ovarian fibromas)
 Support services
Other:

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SWOT ANALYSIS



THREATS Research

Across the country, patients with Gorlin syndrome do not currently received a standardised and equitable service. Their care is not always timely and variation in practice results in over and under treatment and patients being lost to follow up or receiving different levels of care with respect to screening and follow up.

The Manchester Cancer Skin Pathway Board believes that the expansion and formalisation of the multidisciplinary clinic model developed in Manchester would address these problems. We believe that specialist clinics will massively simplify the care pathway and reduce numerous visits to numerous hospitals and numerous specialists with poor co-ordination between them at present.

The Skin Pathway Board will gather evidence to make the case for this expansion to Manchester Cancer and commissioners. This work will also have the potential to inform the development of services elsewhere in the country.

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WHERE ARE WE NOW



Collaboration with Salford Royal, Central Manchester University Hospitals, The Christie and Greater Manchester Cancer.



Funding agreed and provided by Macmillan.



Integrated Research Application System (IRAS) application in process for ethics.



Questionnaire design with appropriate input from Gorlin Community groups, Manchester Skin Care Board & Macmillan.



Project plan with timescales for delivery approved



Awareness and integration into specialist services of Photo Dynamic Therapy, Surgical Intervention, Genetics and Dermatology.

Please welcome Sally Webster

Gorlin Patient & Carer

DISCUSSION

<http://www.gorlingroup.org/>

Experience of living with / supporting someone living with gorlin syndrome.

Navigating the system.

What's important in terms of holistic support?

What do we need?

What would make a difference?

Thank you.