

Gorlin syndrome in the United Kingdom: a survey of patient needs, treatment availability and outcomes

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Background

- Gorlin syndrome, also known as nevoid basal cell carcinoma (BCC) syndrome or basal cell nevus syndrome, is an autosomal dominant genetic disorder¹ with an estimated prevalence in the UK of 1 in 30,000²
- The formation of multiple BCCs and odontogenic keratocysts are the main diagnostic criteria¹
- Other diagnostic criteria include palmar and/or plantar pits, ectopic calcifications of the falx cerebri, congenital skeletal abnormalities, cleft lip and/or palate and eye abnormalities¹
- A number of treatment options are available for the management of BCC in Gorlin syndrome, however, the patient population with Gorlin syndrome in the UK has not been well characterised, with little data available regarding the prevalence of the various symptoms of the disease and the availability of treatment
- The Gorlin Syndrome Group was formed in the UK in 1992 with the aims of supporting patients with Gorlin syndrome and their families, educating medical professionals and promoting research into Gorlin syndrome
- In collaboration with the Gorlin Syndrome Group, we conducted a survey of patients with Gorlin syndrome with the aims of:
 - Improving knowledge of the patient population with Gorlin syndrome in the UK
 - Assessing awareness of Gorlin syndrome amongst healthcare professionals from the perspective of patients
 - Determining the quality-of-life effects of Gorlin syndrome
 - Assessing the availability and uptake of treatment for symptoms of Gorlin syndrome

Methods

- To address the objectives outlined above, a 20 question survey was constructed to assess:
 - The latency between the onset of symptoms and diagnosis of Gorlin syndrome
 - The prevalence and severity of the various symptoms of Gorlin syndrome
 - Awareness of Gorlin syndrome among general practitioners (GPs) and other healthcare professionals
 - The impact of Gorlin syndrome on patient quality of life
 - The availability and uptake of treatment and support services
- The survey was sent to the 243 members of the Gorlin Syndrome Group, and was also made available on the Gorlin Syndrome Group's website and Facebook page to allow access by non-members with Gorlin syndrome in the UK
- Patient-rated severity of Gorlin syndrome was assessed on a 10-point numerical rating scale, with a rating of 1 indicating that the patient had not required treatment and 10 indicating the requirement for multiple treatments every year with a significant impact on the patient's life
- GP and consultant awareness of Gorlin syndrome, as well as the usefulness of information provided by GPs, were rated on a 5-point numerical rating scale, with 1 indicating very poor awareness and 5 indicating very good awareness

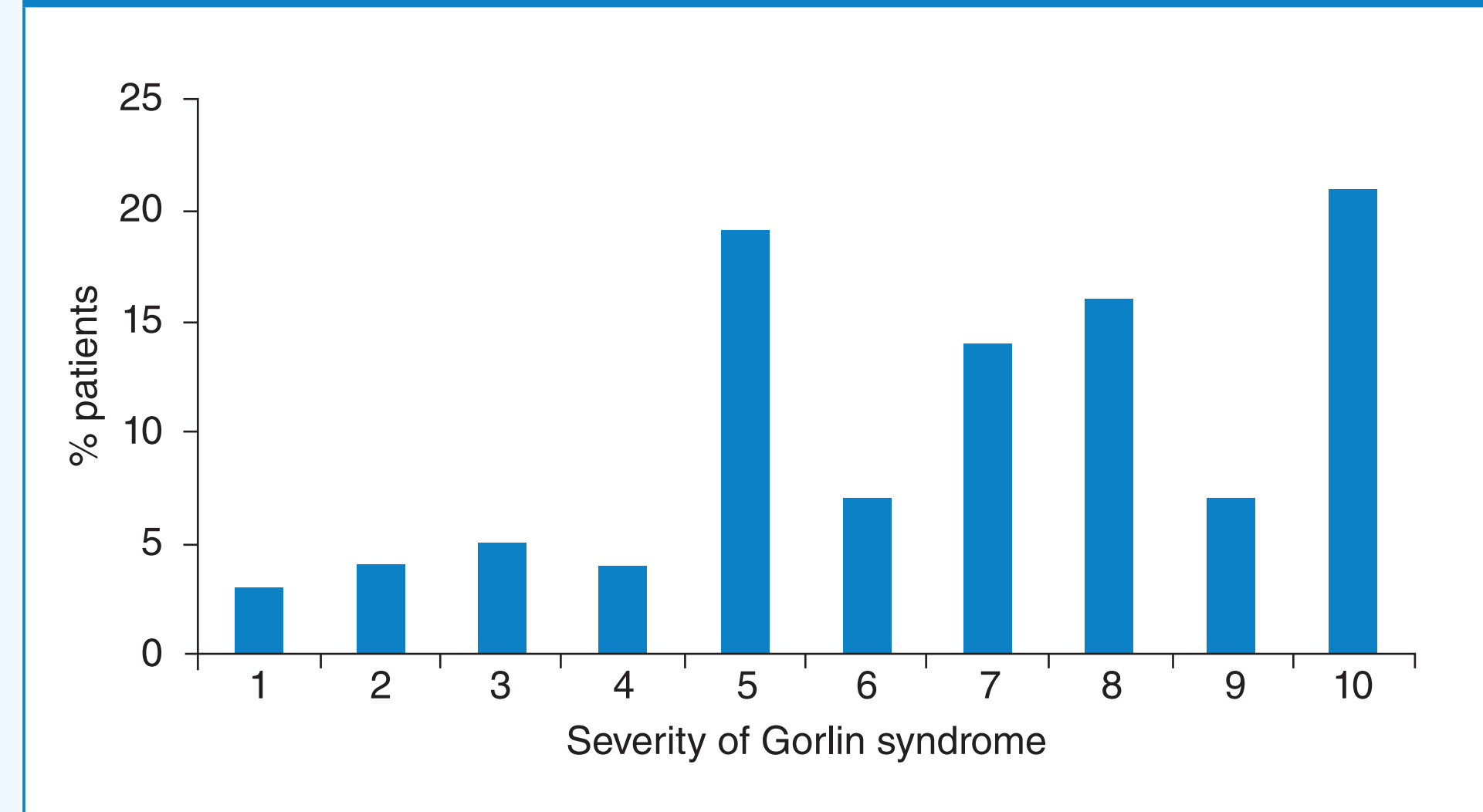
Results

- The survey was conducted over the period from 20 March 2013 to 30 April 2013
- A total of 73 surveys were completed; 67 responses were completed by patients with Gorlin syndrome and 6 responses were completed on behalf of a patient with Gorlin syndrome by a family member or friend

Diagnosis of Gorlin syndrome

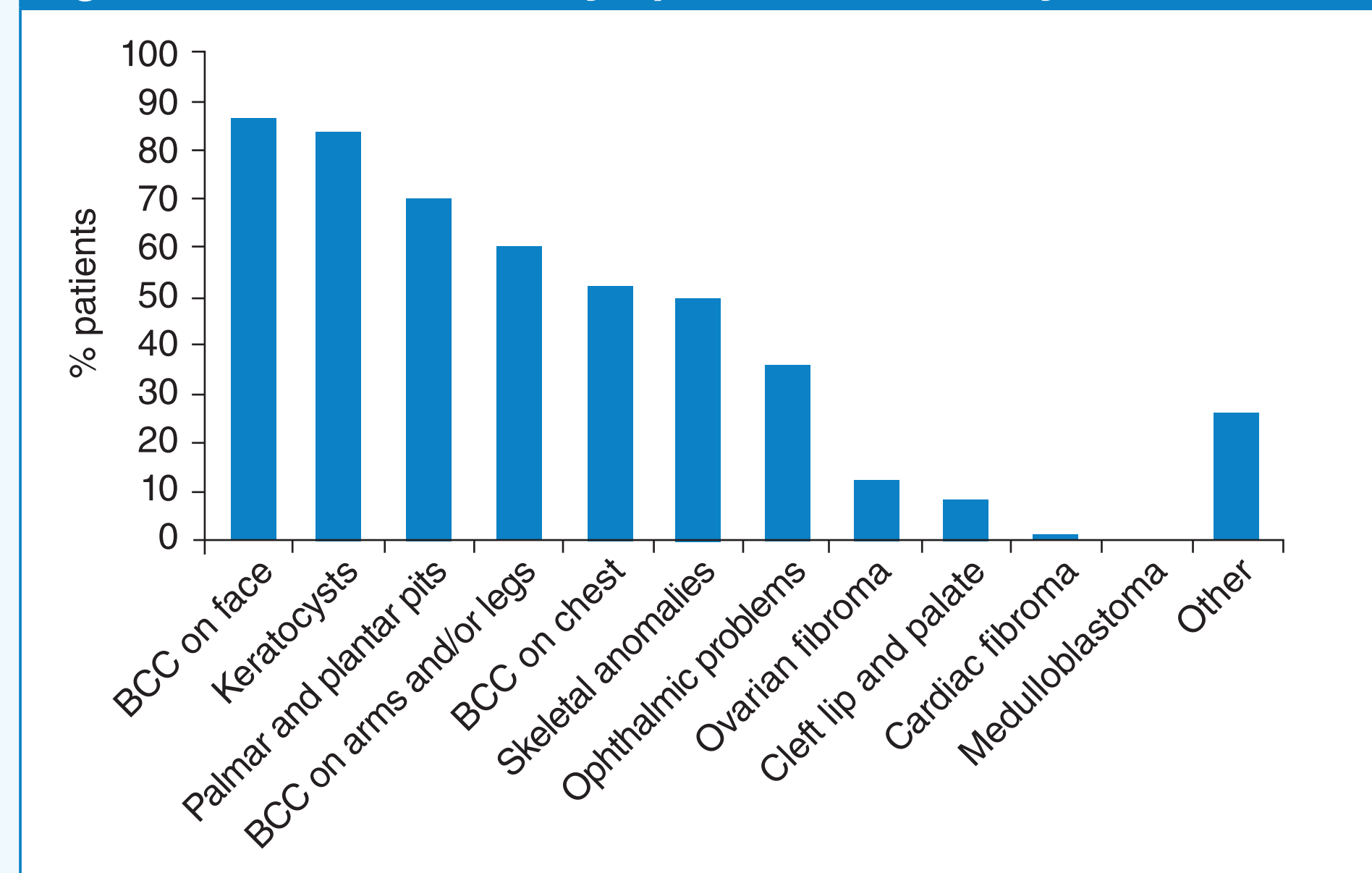
- The median age of patients with Gorlin syndrome was 49 years (range 3–86)
- The median time since diagnosis of Gorlin syndrome was 25 years (range 0–74)
- The median patient-rated disease severity was 7, with 21% of patients rating their disease with the highest severity score of 10 (Figure 1)

Figure 1: Patient-rated severity of Gorlin syndrome



- The median time between onset of symptoms and diagnosis was 4 years (range 0–58)
 - 32% of patients were referred to a specialist or diagnosed immediately at the time of onset of symptoms
 - Although the majority of patients had one consultation with a GP prior to referral, other patients had up to 50 consultations before referral
- Patient ratings of GP awareness of Gorlin syndrome indicated little awareness, with a median rating of 2 (poor); only 38% of patients rated GP awareness as 3 or higher (acceptable, good or very good)
 - The most frequently cited factor affecting the rating of GP awareness was their lack of knowledge of Gorlin syndrome
- In contrast to the ratings of GPs, 73% of patients rated their consultants as having good or very good awareness of Gorlin syndrome
- The prevalence of the various symptoms of Gorlin syndrome experienced by the participants is shown in Figure 2
 - The most frequent symptoms were BCC, keratocysts, palmar and plantar pits, and skeletal anomalies

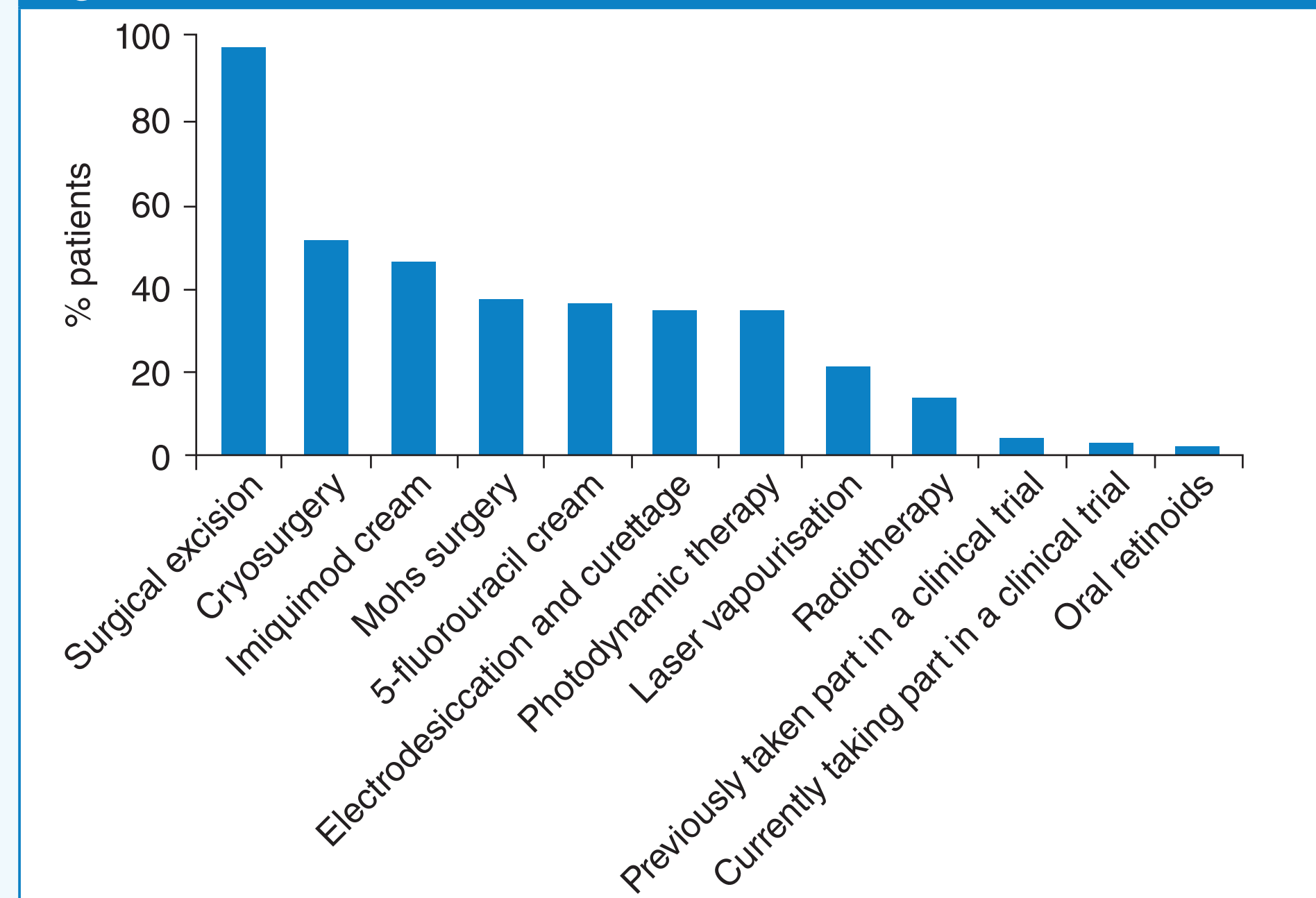
Figure 2: Prevalence of symptoms of Gorlin syndrome



Treatment of Gorlin syndrome

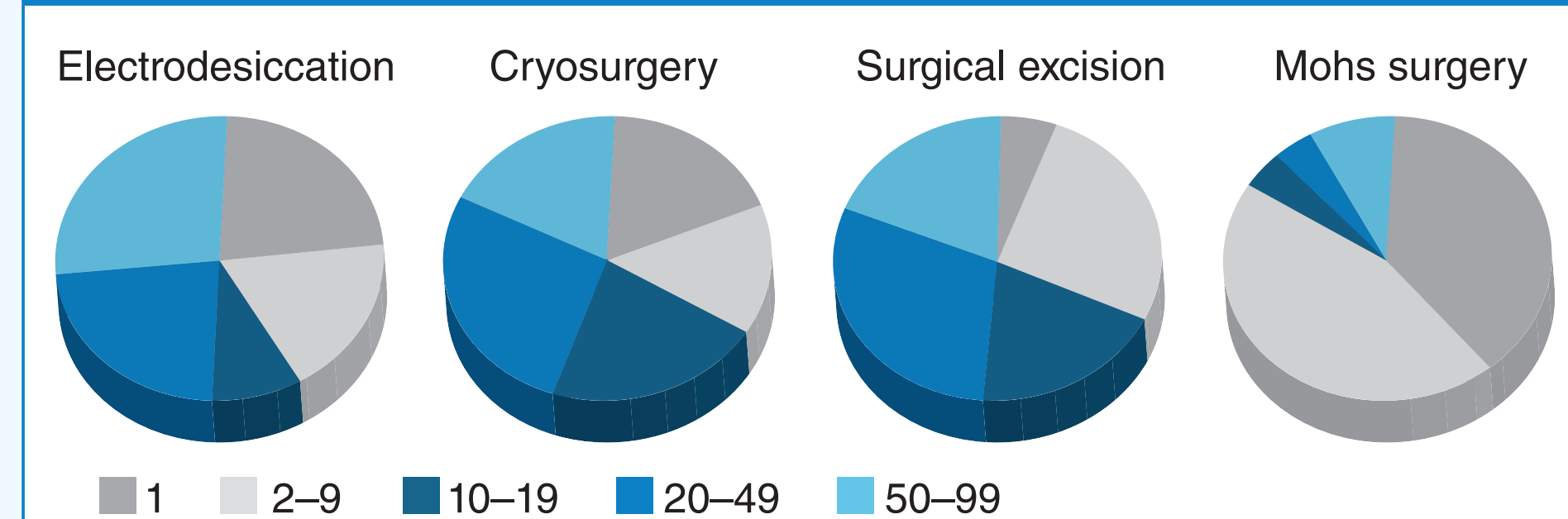
- Treatments received by the survey participants for BCCs are shown in Figure 3
- The vast majority of patients (97%) were treated using surgical excision

Figure 3: Treatment methods for BCC



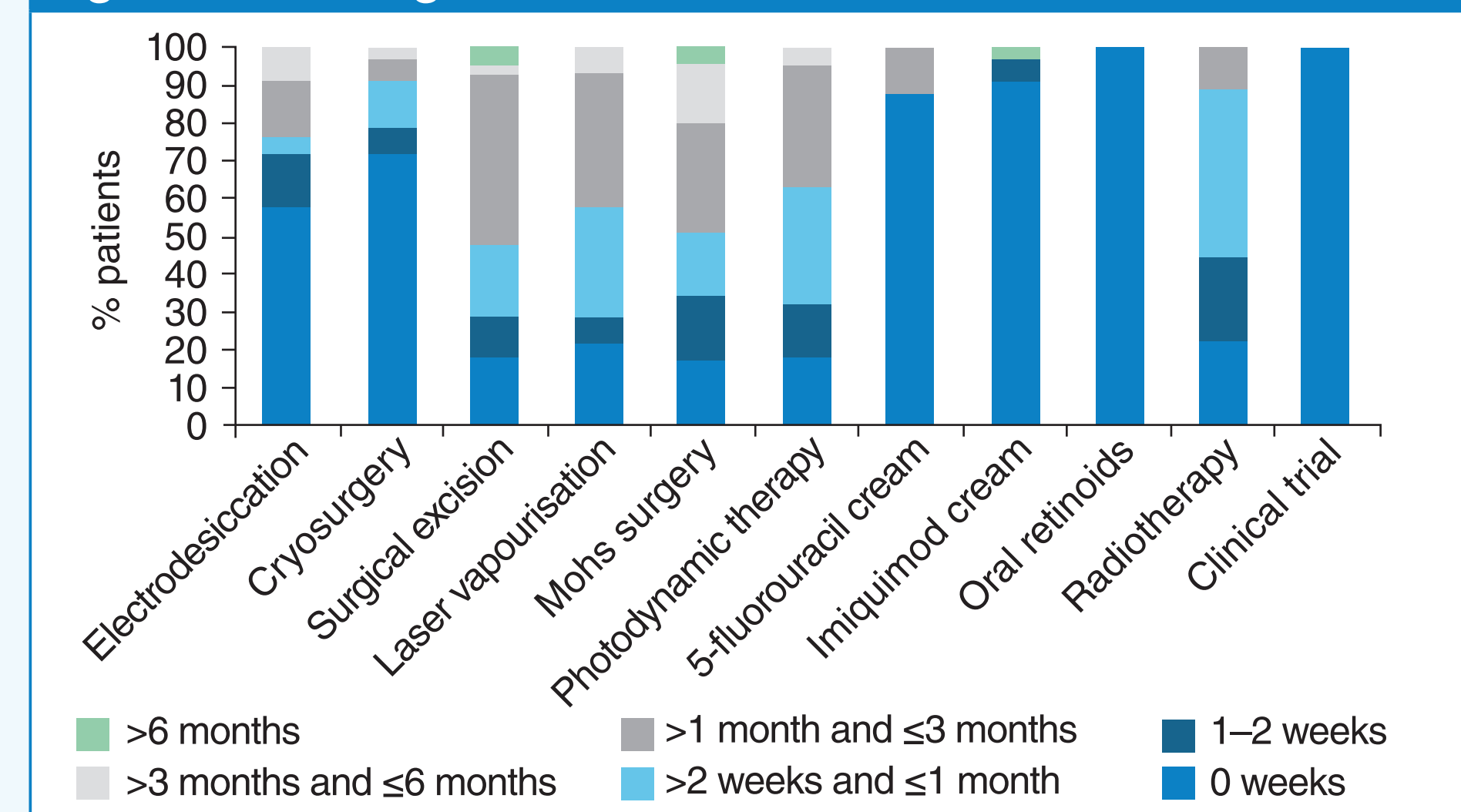
- Study participants generally received more than one type of treatment, with the number of times patients were treated with each treatment modality shown in Figure 4
 - Approximately 50% of patients who had received surgical excision had undergone 20 or more procedures
 - A high number of procedures were also reported by patients receiving electrodesiccation and cryosurgery
 - More than half of patients undergoing surgical excision, cryosurgery or electrodesiccation received treatment for BCCs once every few months
 - Approximately 85% of patients requiring surgical excision of BCCs underwent surgery at least once yearly

Figure 4: Number of electrodesiccation, cryosurgery, surgical excision and Mohs surgery procedures performed



- Waiting times for various methods of treatment for BCC are shown in Figure 5
 - 7% of patients had a waiting time of over 3 months for surgical excision

Figure 5: Waiting times for BCC treatment



- The healthcare professionals consulted regarding the treatment of Gorlin syndrome and frequency of consultation are shown in Table 1
 - Patients who consulted with dermatologists and specialist nurses had the highest frequency of consultations
 - Only 15% of patients consulted with a multidisciplinary team
 - The median distance travelled for consultations was generally low (≤11 miles), but some patients travelled much further

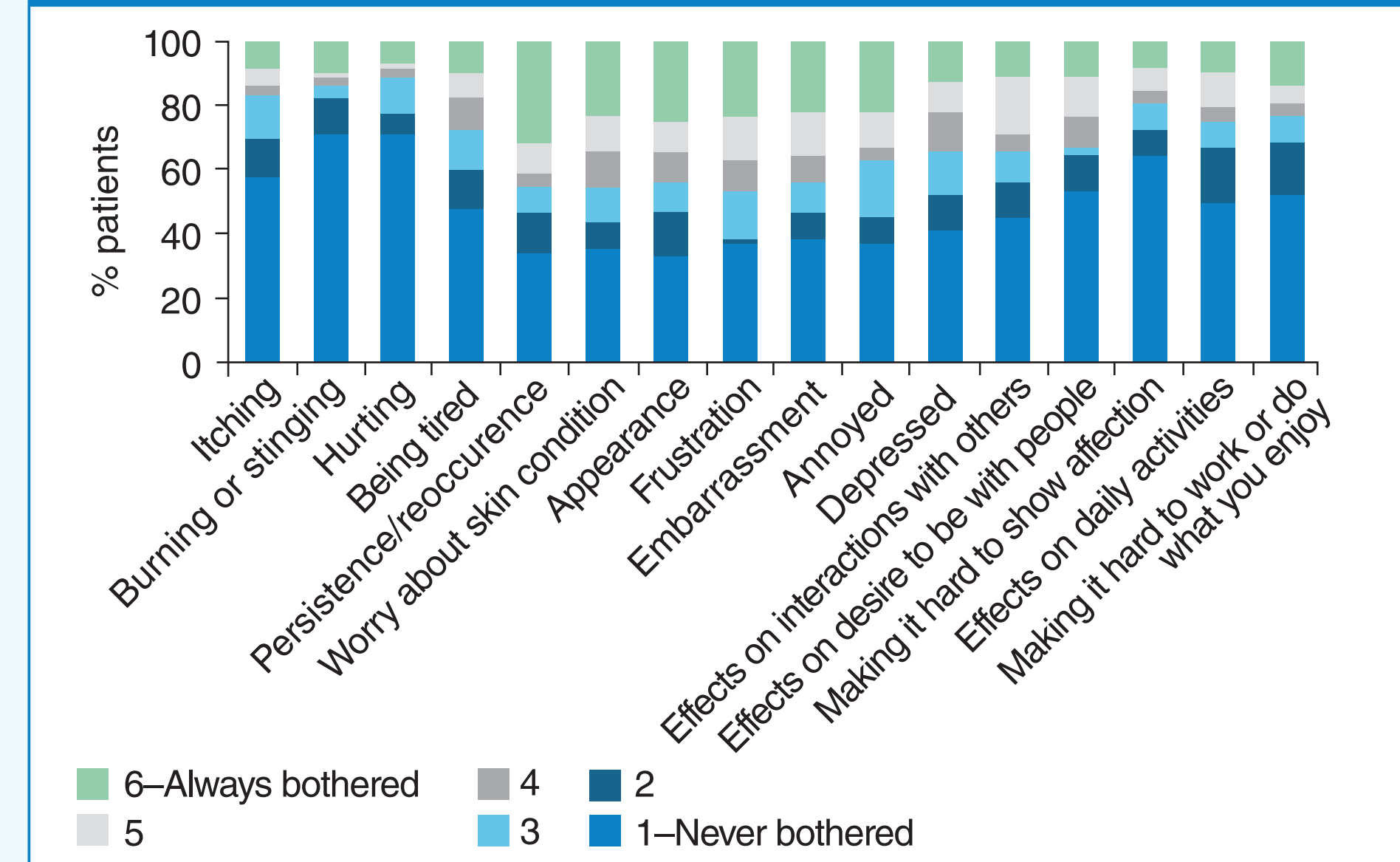
Table 1: Proportion of patients consulting with each type of healthcare professional, frequency of consultation, and distance travelled to consultations

Healthcare professional	Proportion of patients (%)	Frequency of consultation (%)				Median distance travelled, miles (range)
		1-2 months	3-6 months	12 months	2 years	
Dermatologist	74	9	76	16	0	6 (0-99)
Maxillofacial surgeon	56	0	32	47	21	8 (1-99)
Plastic surgeon	43	12	50	27	12	11 (0-99)
GP	34	10	57	19	14	1 (0-5)
Specialist nurse	18	0	82	18	0	3 (0-50)
Multidisciplinary team	15	0	44	22	33	3 (0-37)
Gynaecologist	5	33	33	33	0	10 (3-35)
Other	23	0	29	43	29	10 (0-99)
Not currently seeing anyone	3					

Impact of Gorlin syndrome on quality of life

- Undergoing multiple surgeries had a significant emotional impact on 49% of patients, and had a modest emotional impact on 25% of patients
- The most disliked aspects of treatment for Gorlin syndrome were discomfort (62%), pain (55%), having to take time off work (40%), treatment success rate (29%), dressings (27%) and poor access to treatment (11%)
- Approximately 80% of survey respondents had not been offered counselling as part of their treatment for Gorlin syndrome
- Patient ratings of how much they are bothered by Gorlin syndrome-related skin problems are shown in Figure 6
 - The most common reasons for skin problems bothering patients were persistence/reoccurrence, worry, appearance, frustration and embarrassment

Figure 6: Patient ratings of how much they are bothered by skin-related problems



Conclusions

- Treatments for BCCs in patients with Gorlin syndrome are varied, but the most common is surgical excision
- Multiple surgeries are often required, with over half the patients surveyed requiring a procedure every few months, which has a significant impact on quality of life
- The results of the survey indicate that patients feel GPs have a poor knowledge of the disease, and there is often a significant delay between onset of symptoms and diagnosis
- Once referred, the patients surveyed indicated that the consultant generally had a good understanding of the disease