

Bardet-Biedl Syndrome Patient Journey



Bardet-Biedl Syndrome UK
1027384 & SCO41839
www.bbsuk.org.uk



Pre-Diagnosis

Symptoms:

- Extra Digits
- Obesity
- Delayed Development
- Visual Impairment
- Renal Abnormalities
- Emotional/Learning Difficulties

Referral to Paediatrician and/or Geneticist:

Waiting for appointments...
Waiting for results...

Implications

- Lack of understanding locally
- Relationships break down
- Social isolation
- Financial pressures
- Child struggling at nursery/school/college

Whole family support needed!

Just an over anxious mum!

You're worrying about nothing!

Diagnosis

Realisation & Implications:

- No cure
- Full knowledge of symptoms
- Uncertain prognosis
- Family planning

Issues

- Lack of understanding and knowledge locally
- Appointments overwhelming

Emotions
Anxiety
Fear
Isolation
Stress

Referral to BBS Clinics

Multi-Disciplinary Highly Specialised Service

Bardet-Biedl Syndrome UK

- BBS UK Support Officers
- Clinic Support
- Booklets/Newsletters
- Annual Family Conference
- Activities & Events

"I got in touch with two wonderful people from the BBS charity. They were a huge source of support to me and put me in contact with even more families."

Emotions
Relief
Guilt
Uncertainty
Grief

Treatment

Bardet-Biedl Syndrome Clinics

18 monthly review available across four centres:

- Great Ormond Street Hospital
- Guys & St Thomas' Trust
- Birmingham Children's Hospital
- Queen Elizabeth Hospital, Birmingham

Patient centred one-stop service:

- Geneticist
- Clinical Psychologist
- Endocrinologist
- Dietitian
- Nephrologist
- Speech & Language Therapist
- Ophthalmic Review
- Clinical Nurse Specialist
- BBS UK Support Officer
- DNA testing

Emotions
Anxiety
Stress
Depression
Acceptance

"The care we have received at the clinic has been exceptional. Each clinician has been thorough and explained the condition and diagnosis with great care and detail... we left the clinic with a much greater knowledge of the condition."

Future/Management

Clinical Care

Continued care at a BBS Clinic

Support/Coordination

BBS UK Support Officer:

- Emotional support
- BBS Clinic Liaison Service
- LA referrals
- Benefits information/support
- Advocacy & signposting

Ongoing Challenges:

- Local authority cutbacks across education, health and social care
- Benefit reforms
- Gaining employment
- Gaining independence
- Fighting social isolation

Hopes for Future:

- Continued specialised healthcare provision
- Improved knowledge and understanding of BBS
- Increased research and study of BBS
- Therapies, treatments, technologies

