The UK Bloom syndrome National Clinical Network - An approach to specialist care for BS affected families

Dr Shehla Mohammed
Dr Stefan Meyer
Dr Rui Santos
Mrs Leza Whitman

Blooms Clinical Network in the UK - Some Background

Overview of Clinics
St Thomas' and Manchester

Rare Disease Clinical Network
2023

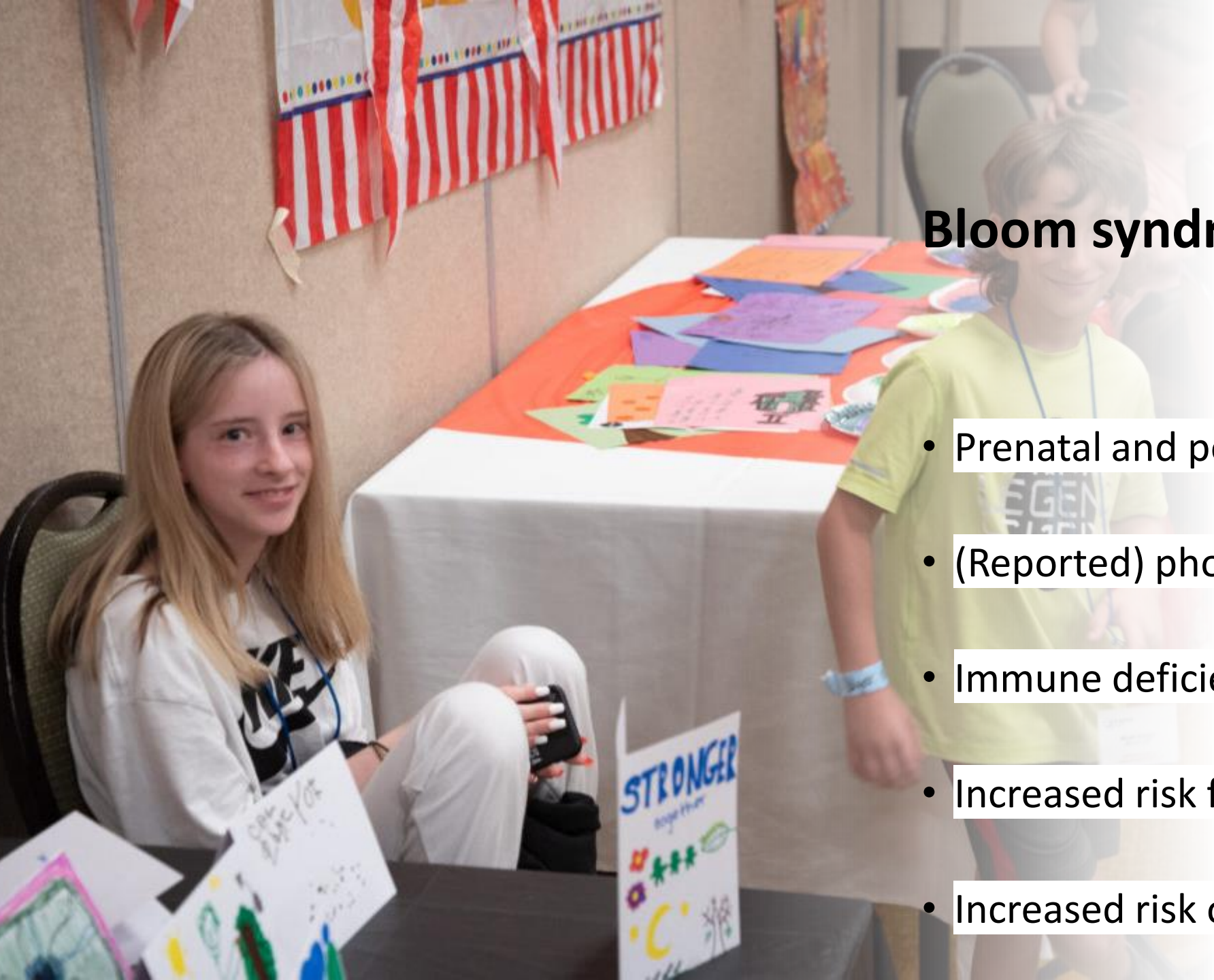
NHS
Bloom's syndrome
Rare Disease
Collaborative Network

Specialist teams & assessments
Nursing input & support

Developing Clinical Pathways

Progress
Observations
Challenges

Family Feedback

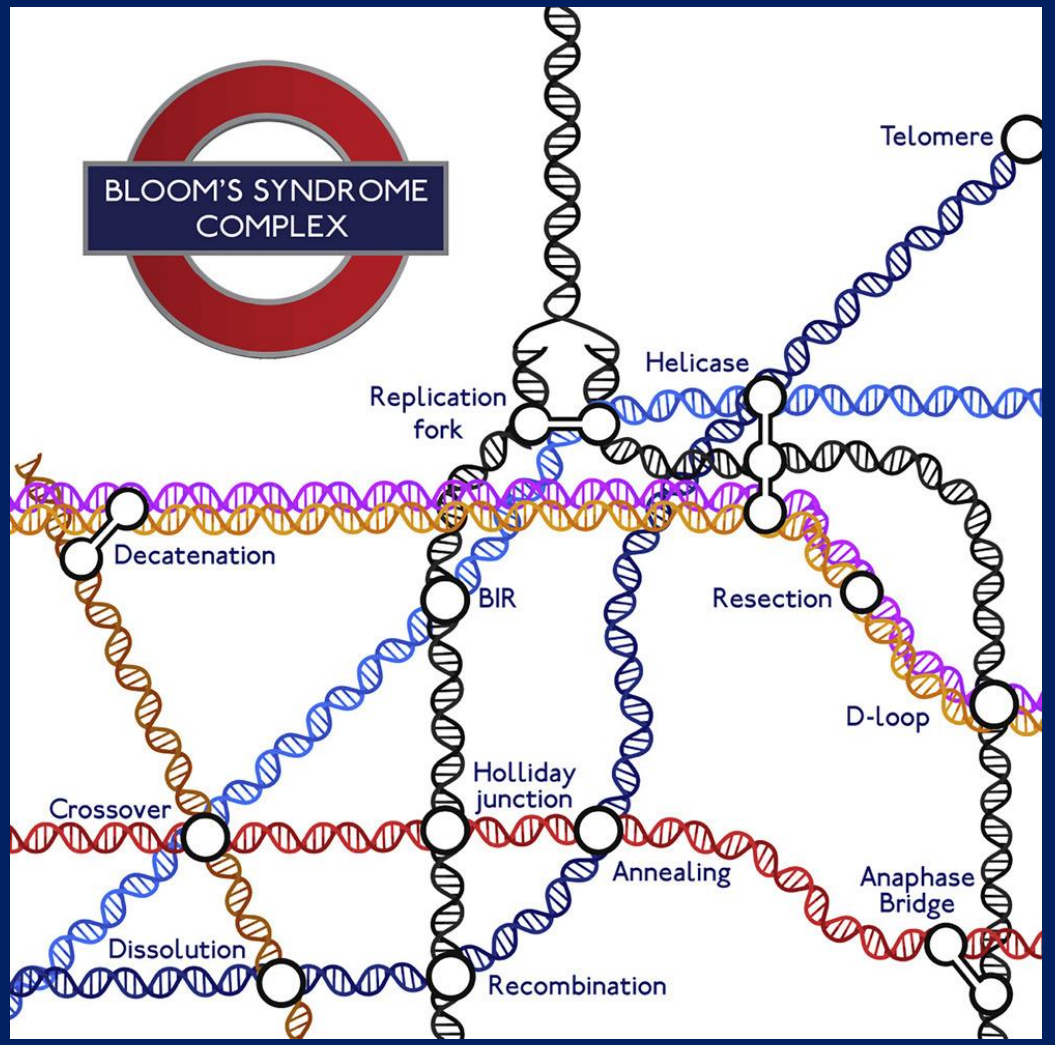


Bloom syndrome –

- Prenatal and postnatal growth deficiency
- (Reported) photosensitive skin changes
- Immune deficiency
- Increased risk for diabetes
- Increased risk of early onset of cancer

Bloom Syndrome

- Multiple system involvement
- Genetic disruption of a fundamental complex maintaining chromosomal stability
- Mostly *BLM* (>90%), rarely *TOP3A*, *RMI1* and *RMI2*



Caretakers of the Genome

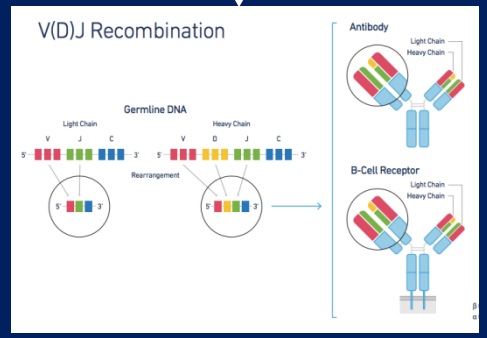
Bloom syndrome

BLM gene

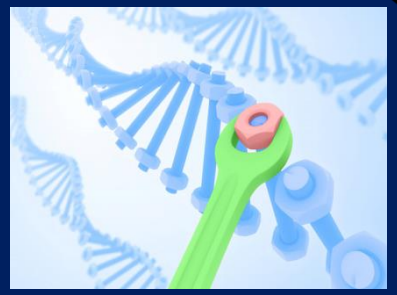
RecQL3

DNA replication

Growth deficiencies
IUGR
Short stature



DNA recombination



DNA repair

Cancer
Leukaemia
Skin cancer

Immunity
Infections
COPD

Centers of Excellence for Rare Diseases - purpose

- Deliver high quality coordinated care
- Promoting information and support for families
- Adequate caseload for expertise
- Not dependent on a single clinician
- Transition of care from children's to adult services

Guy's and St Thomas' NHS
NHS Foundation Trust

Developing MDT space

- RDC space
- Old phototherapy unit
- Victorian part of St Thomas’
- Some structural elements to work around

- Designed and built in partnership with families and patients with specialist needs
- National Highly specialist Clinics only
- Designated for Children, adults and transition care

Guy's and St Thomas' NHS
NHS Foundation Trust

Guy's and St Thomas' NHS
NHS Foundation Trust

Centers of Excellence

- Education and training
- Research active
- Membership of international networks of excellence

Engaged with patients and families with rare /ultra rare diseases

Blooms Service provision – where did we start ?

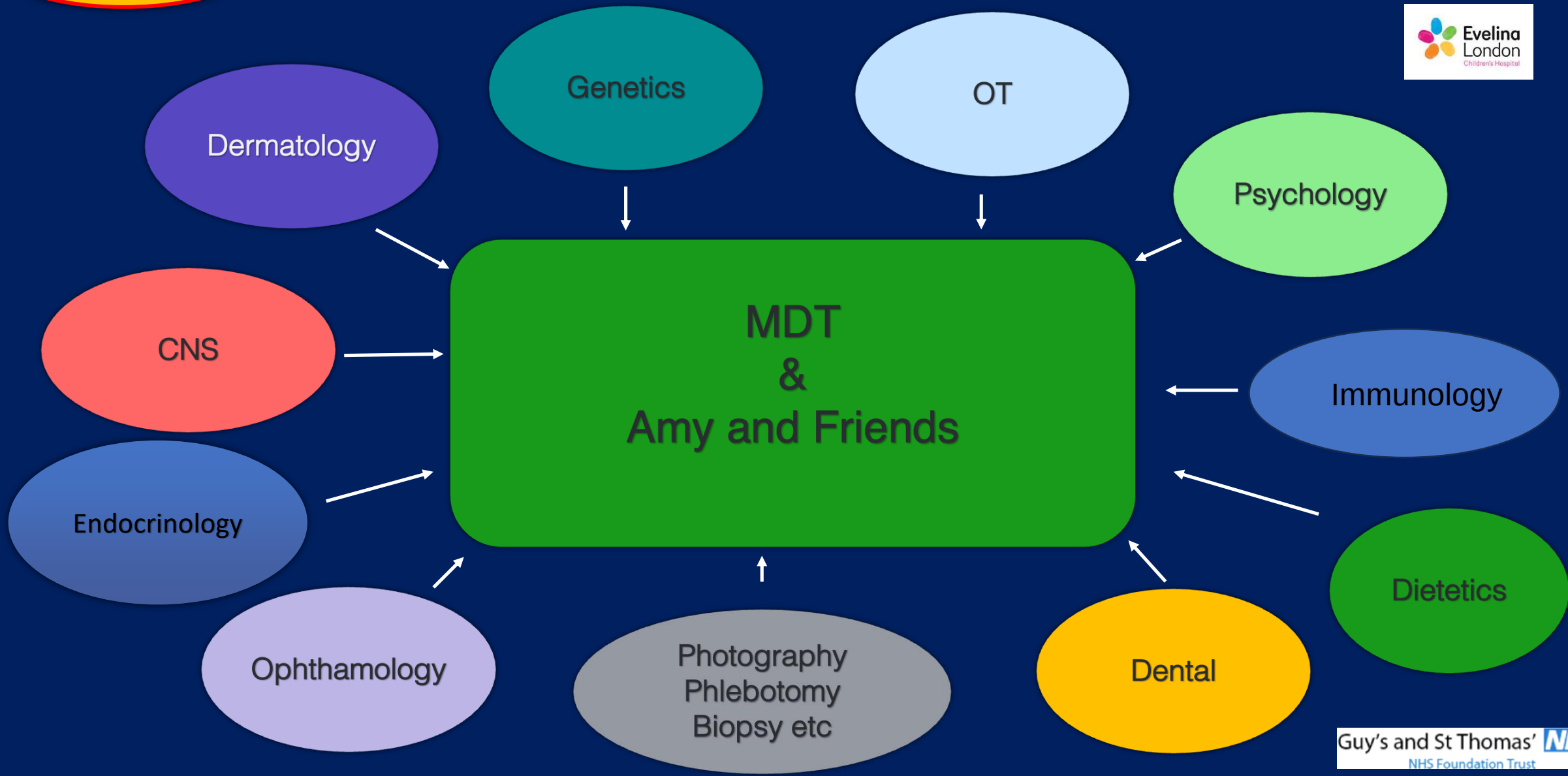
- Meet complex clinical needs
- Equitable access to care
- Agreed care pathways
- Need to monitor and tailor individual management needs
- Liaise with local services for ongoing management

Patient-centred clinical model

Guy's and St Thomas' NHS
NHS Foundation Trust

FORMAT OF CLINICS FOR BLOOMS SYNDROME

50+ appointments



Evelina London Children's Hospital

Guy's and St Thomas' NHS
NHS Foundation Trust

National out-reach nursing : home visits

- Complexity and co-morbidities of Bloom’s syndrome
- Detailed nursing assessment
- Assessing child in home environment
- Formulating acute care plan and care passport
- Care passport is a dynamic document
- Advanced Care Plan

Paula

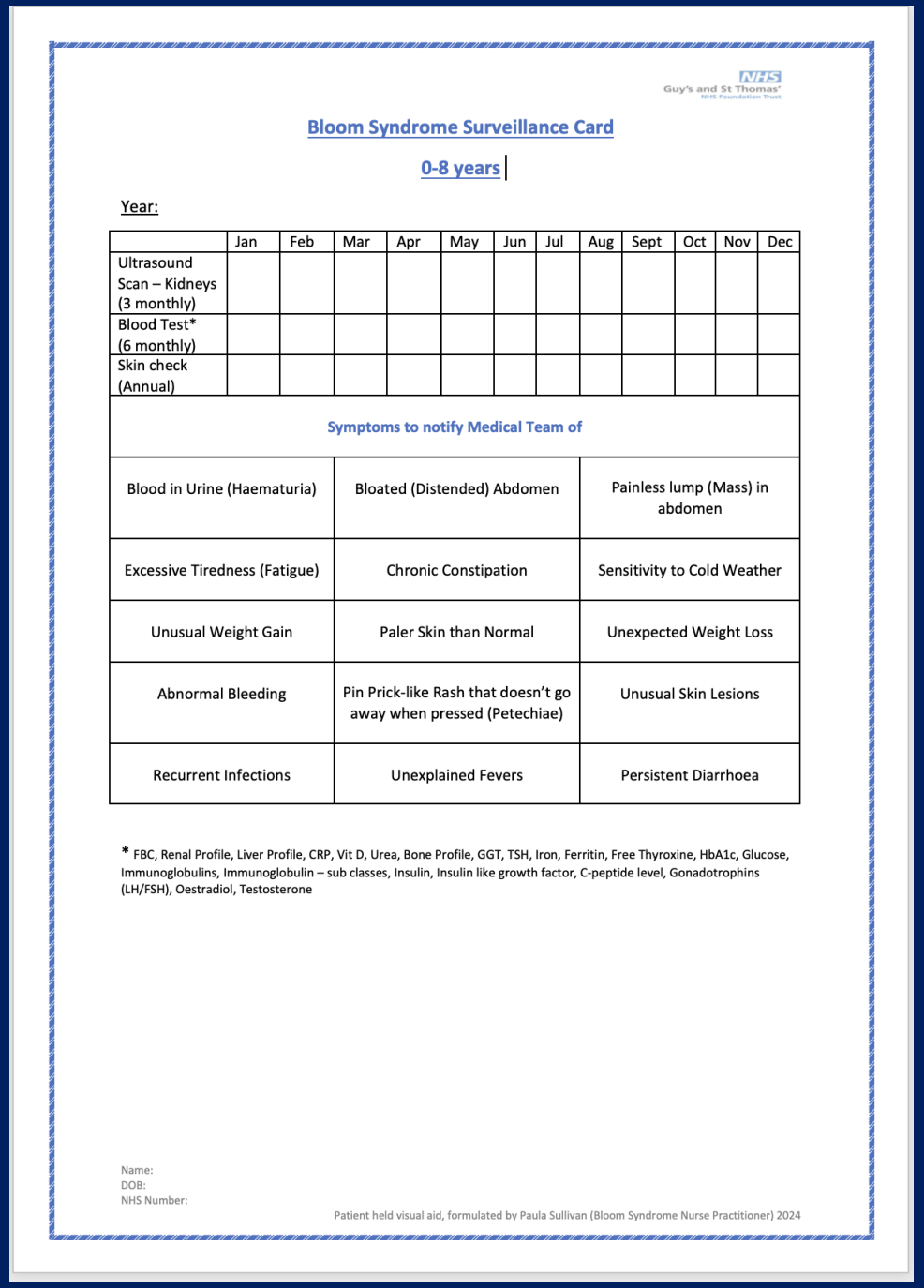
Phillipa

Good practice /Innovation

- Bespoke management plan, shared with family and local teams

My Blooms Syndrome Care passport

- Highlights the child's individual needs and allows professionals to be aware of each child's "normal" and how to care for them as their family would. It also ensures that the child has a voice even when they are unable to articulate their wants and needs.
- IP arrangement between GSTT and "Amy and Friends" to allow them to share the passport internationally with families that need it



Guy's and St Thomas' NHS Foundation Trust

Bloom Syndrome Surveillance Card

0-8 years

Year:

	Jan	Feb	Mar	Apr	May	Jun	Jul	Aug	Sept	Oct	Nov	Dec
Ultrasound Scan – Kidneys (3 monthly)												
Blood Test* (6 monthly)												
Skin check (Annual)												

Symptoms to notify Medical Team of

Blood in Urine (Haematuria)	Bloated (Distended) Abdomen	Painless lump (Mass) in abdomen
Excessive Tiredness (Fatigue)	Chronic Constipation	Sensitivity to Cold Weather
Unusual Weight Gain	Paler Skin than Normal	Unexpected Weight Loss
Abnormal Bleeding	Pin Prick-like Rash that doesn't go away when pressed (Petechiae)	Unusual Skin Lesions
Recurrent Infections	Unexplained Fevers	Persistent Diarrhoea

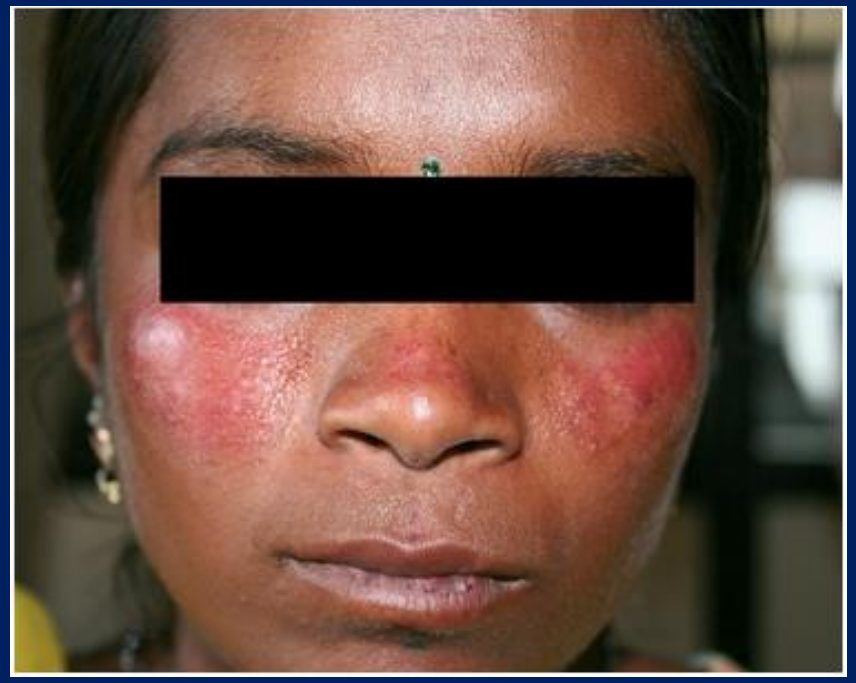
* FBC, Renal Profile, Liver Profile, CRP, Vit D, Urea, Bone Profile, GGT, TSH, Iron, Ferritin, Free Thyroxine, HbA1c, Glucose, Immunoglobulins, Immunoglobulin – sub classes, Insulin, Insulin like growth factor, C-peptide level, Gonadotrophins (LH/FSH), Oestradiol, Testosterone

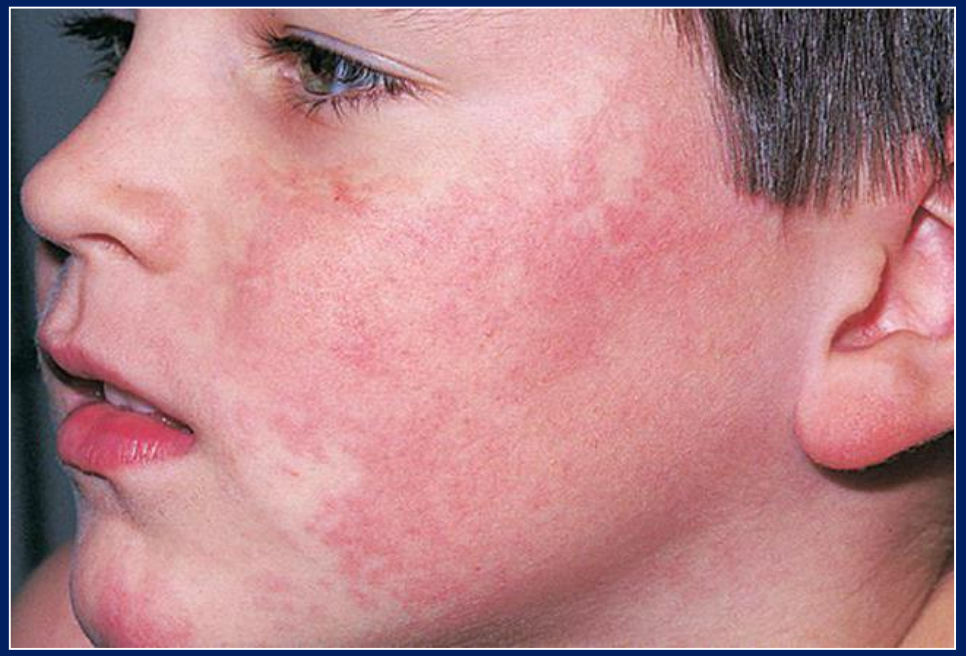
Name:
DOB:
NHS Number:

Patient held visual aid, formulated by Paula Sullivan (Bloom Syndrome Nurse Practitioner) 2024

Surveillance card
Patient held visual aid

Photosensitivity in Bloom syndrome in the literature

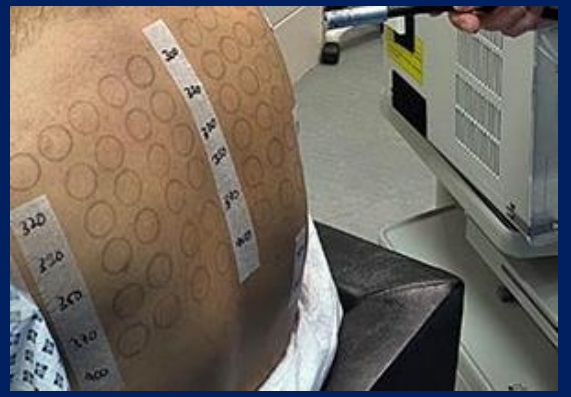


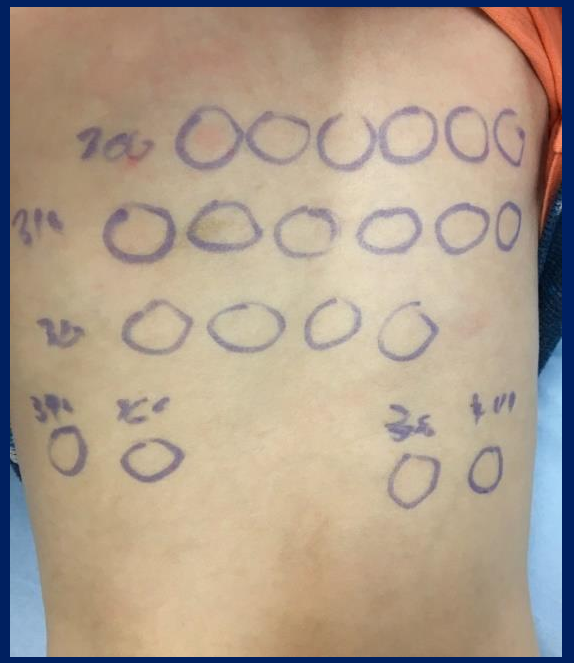


Sultan SJ, Sultan ST. Bloom syndrome in two siblings. Pediatr Dermatol. 2010 Mar-Apr;27(2):174-7; Shah H, Sheth FJ, Pandit VS, Langanecha B. Bloom syndrome: report of two cases in siblings. Int J Dermatol. 2013 Aug;52(8):990-2.

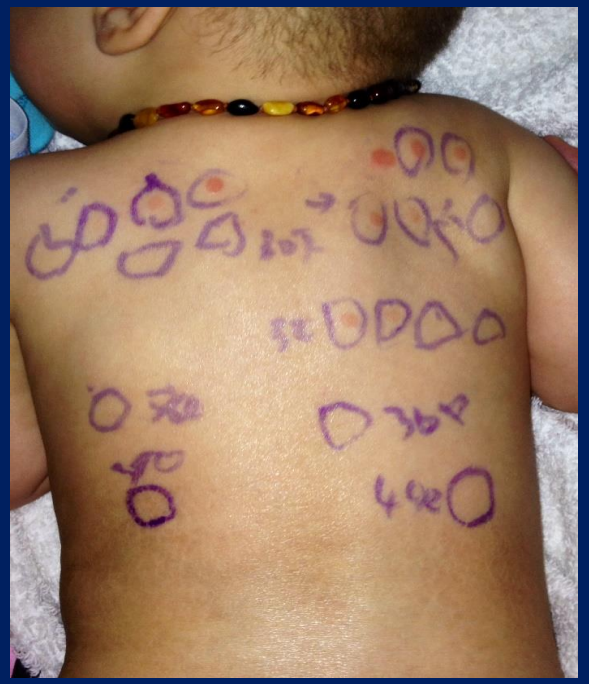
Our study

- All patients were assessed by a photo-dermatologist
- Underwent phototesting to objectively assess photosensitivity
- **8 children: 5 male, 3 female; age range 7-16 yrs**
- Confirmed diagnosis of Bloom syndrome based on abnormal SCE and detection of pathogenic variants in the *BLM* gene

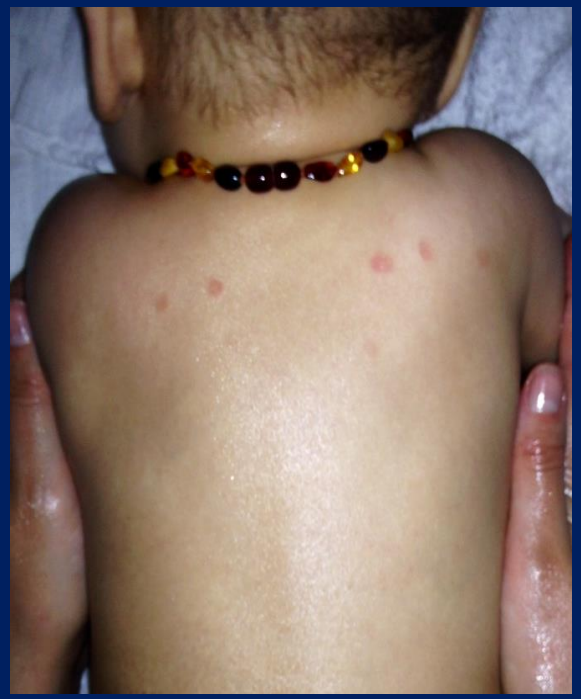




Photosensitivity in Trichothiodystrophy



48 hours



2 weeks

Conclusions

- Series of 8 cases of Bloom syndrome without the characteristic finding of photosensitivity
- Supported by normal phototesting in 7 patients
- First time that photosensitivity in Bloom syndrome has been objectively assessed
- Acute photosensitivity is not a cardinal feature of Bloom syndrome
- Increased risk of non-melanoma skin cancer from early 30s remains - regular skin surveillance and sun protection measures vital
- Bloom syndrome long term natural history study planned

Imaging in Bloom Syndrome

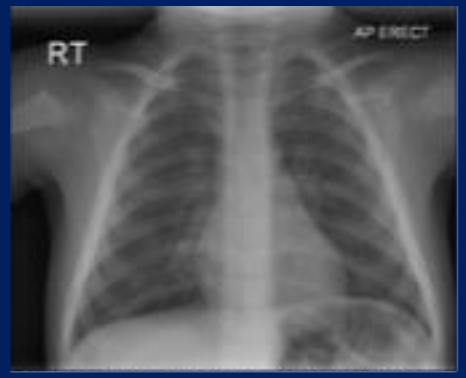
X-rays

Ultrasound

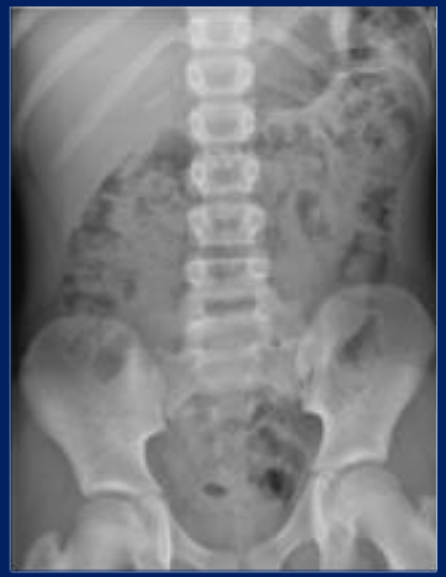
CT/MRI

Other scans

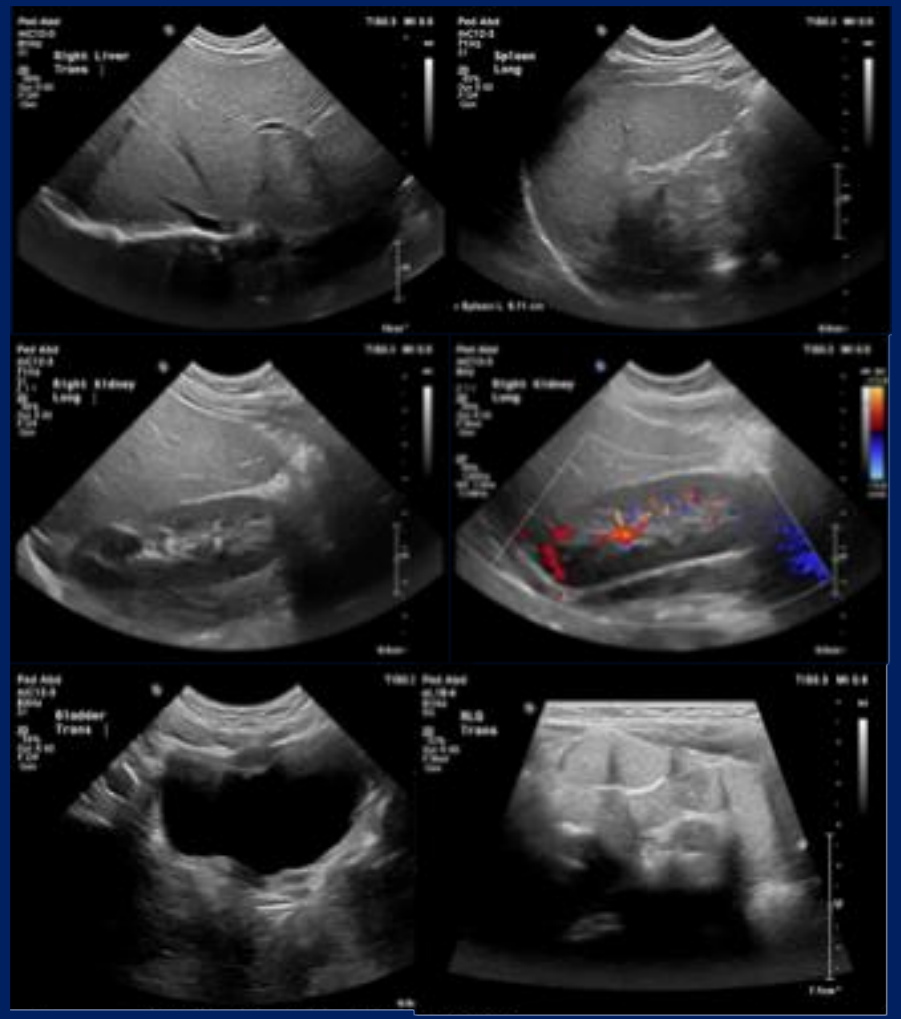
Imaging in Bloom Syndrome



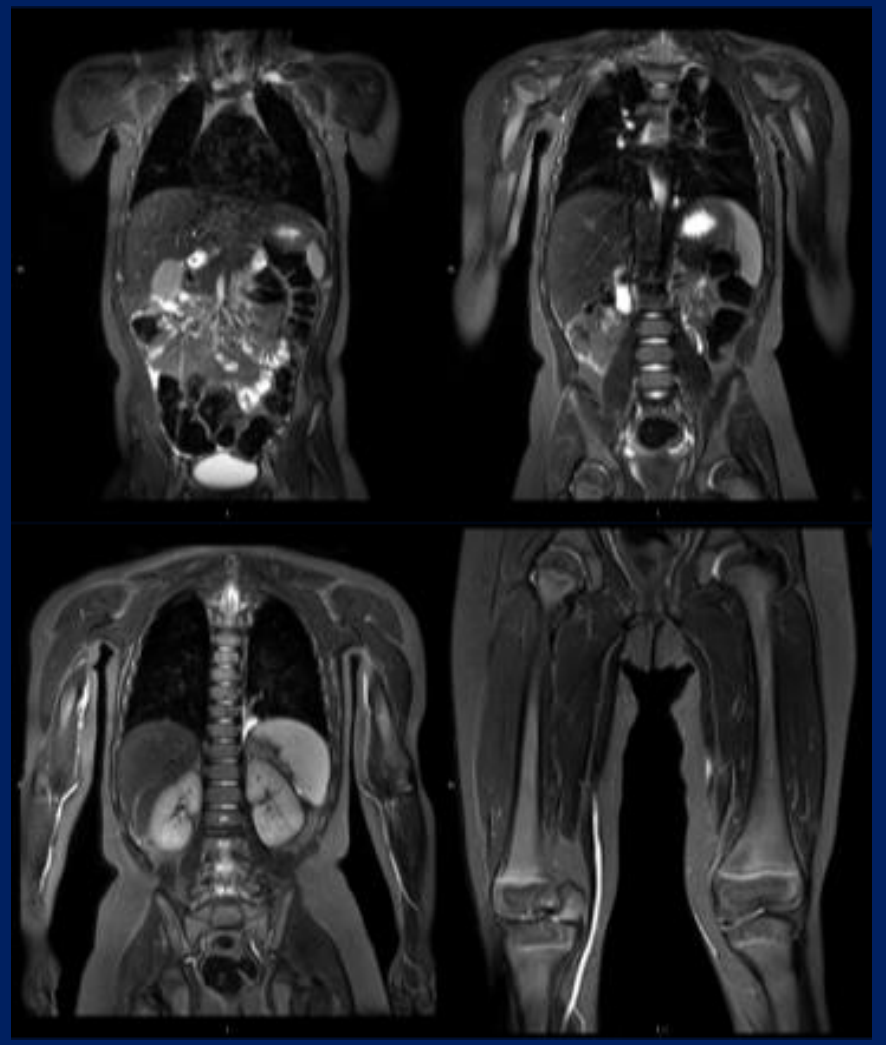
Case courtesy of Ian Bickle, Radiopaedia.org, rID: 46487



Case courtesy of Ian Bickle, Radiopaedia.org, rID: 46488

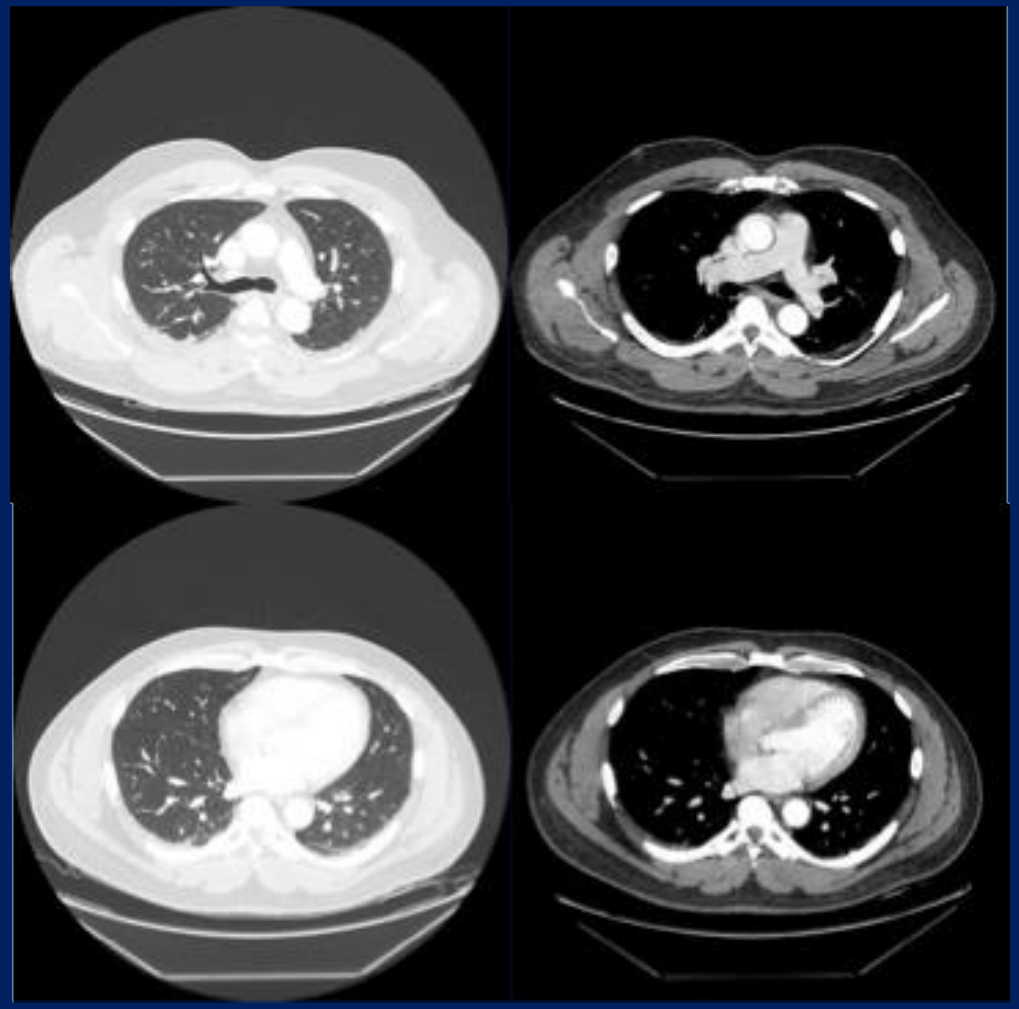


Case courtesy of Ryan Thibodeau, Radiopaedia.org, rID: 166684

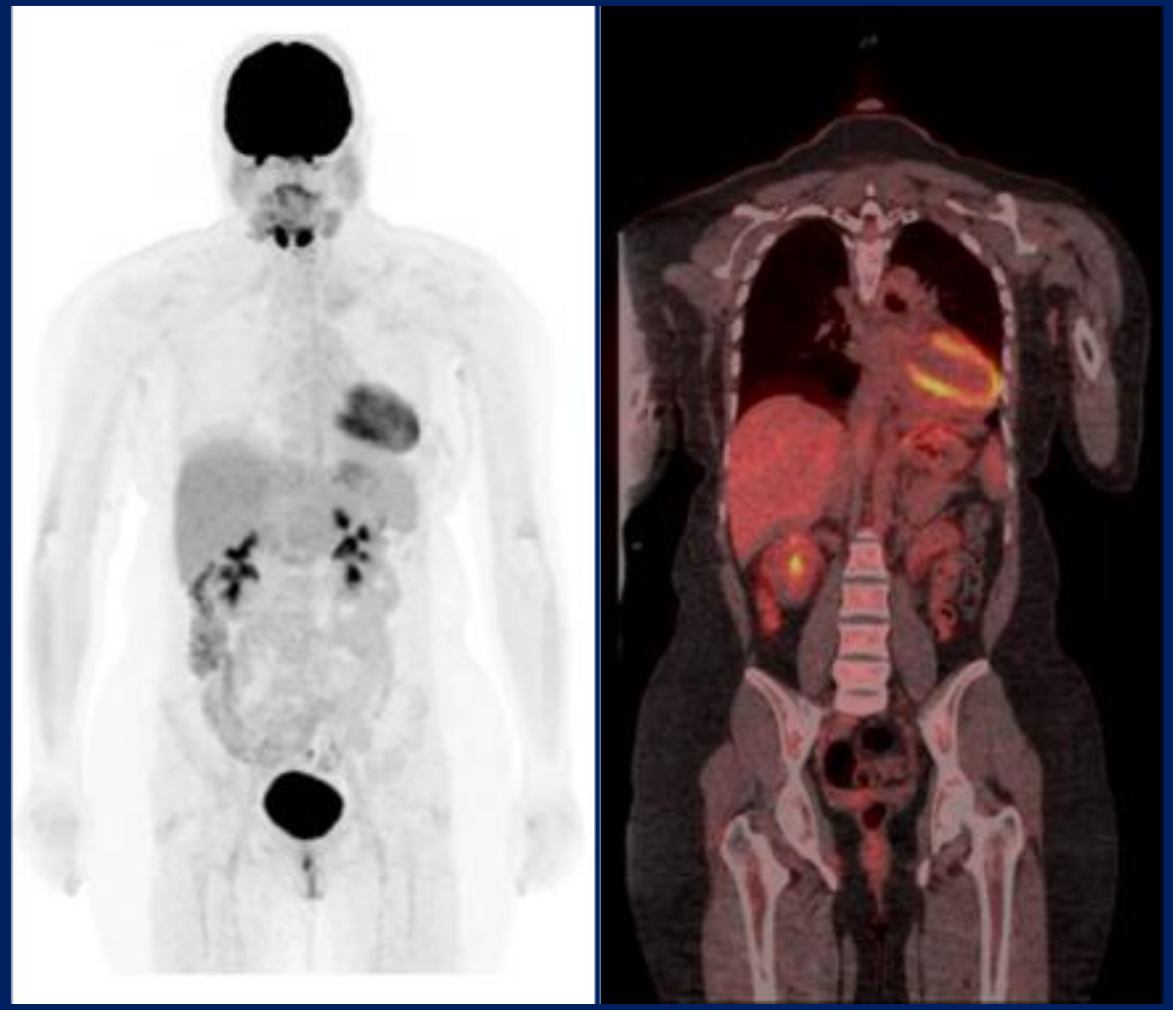


Case courtesy of Jeremy Jones, Radiopaedia.org, rID: 100224

Imaging in Bloom Syndrome



Case courtesy of Stefan Tigges, Radiopaedia.org, rID: 98460



Case courtesy of David Little, Radiopaedia.org, rID: 77382

Blooms Service provision – where did we start and where are we now

- Patients with BS
  - Manchester 10 (8)
  - London 10

- Genetics 16/18
- Regular reviews – imaging, MDT, other specialities partly via zoom/teams

- Skin/photosensitivity study – London …patients

- Ongoing project : imaging of BS

AIMS OF THE SERVICE

- Provide focused care for individuals with BS (…*we are interested*!)
- Address medical screening and prevention needs
- Coordinate multidisciplinary input (imaging, surgeons, endocrine, fertility etc.)
- Study the long-term health implications of BS
- Understand genotype/phenotype aspects and genetic drivers
- Collect clinical and biological data on BS-associated pre-cancer and cancer
- Collect clinical data on therapeutic interventions (cancer and non-cancer)

Enabling translational work - (…..this is a beginning……)

Ongoing collaborative work with

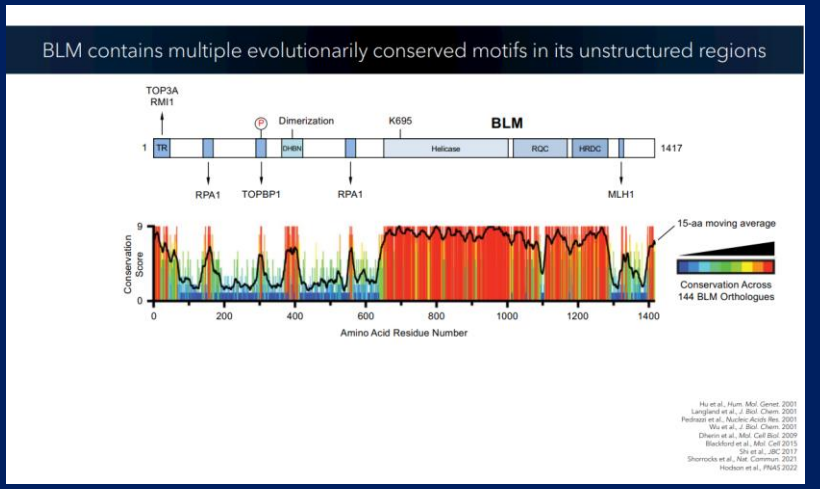
A Blackford - Oxford and Copenhagen (BS protein function)

C Schmidt - Manchester (BS protein function)

R Bowden – Cambridge (genetics, germline and acquired)

C Cunniff and N Kucine -(Registry)

V Chang – UCLA – (screening and prevention, translational studies)



National Bloom Syndrome Clinical Team

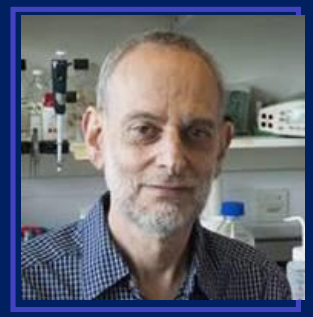
Dr Shehla Mohammed
Service Lead
Consultant Clinical Geneticist

Dr Mike Harrison
Consultant Paediatric Dentist

Dr Hiva Fassihi
Consultant Dermatologist

Dr Stefan Meyer
Consultant Oncologist
National Lead for Blooms Service

Ms Paula Sullivan
Nurse Practitioner



Professor Alan Lehmann
Honorary Consultant Scientist
University of Sussex

Sonia Cardozo
Assistant Service Manager

Ms Susie Morley
Consultant
Ophthalmologist and Oculoplastic surgeon

Ms Julia Hopkins
Paediatric Dietitian

Ms Katy Strudwick
Paediatric Occupational
Therapist

Dr Jennifer Baulcomb
Neuropsychologist &
Educational Psychologist

Phillipa Sellar
Nurse Practitioner

Dr Rui Santos
Consultant Radiologist

Dr Ian Kesterton
Clinical Scientist

Dr Yvonne Rooney
Adult special Care Dentist

Dr Emma McGibbon
Clinical Psychologist

Dr Ramsay Bowden
Consultant Clinical Geneticist
Assistant Professor
Research

AMY and FRIENDS
COCKAYNE SYNDROME/TRICHOTHIODYSTROPHY SUPPORT

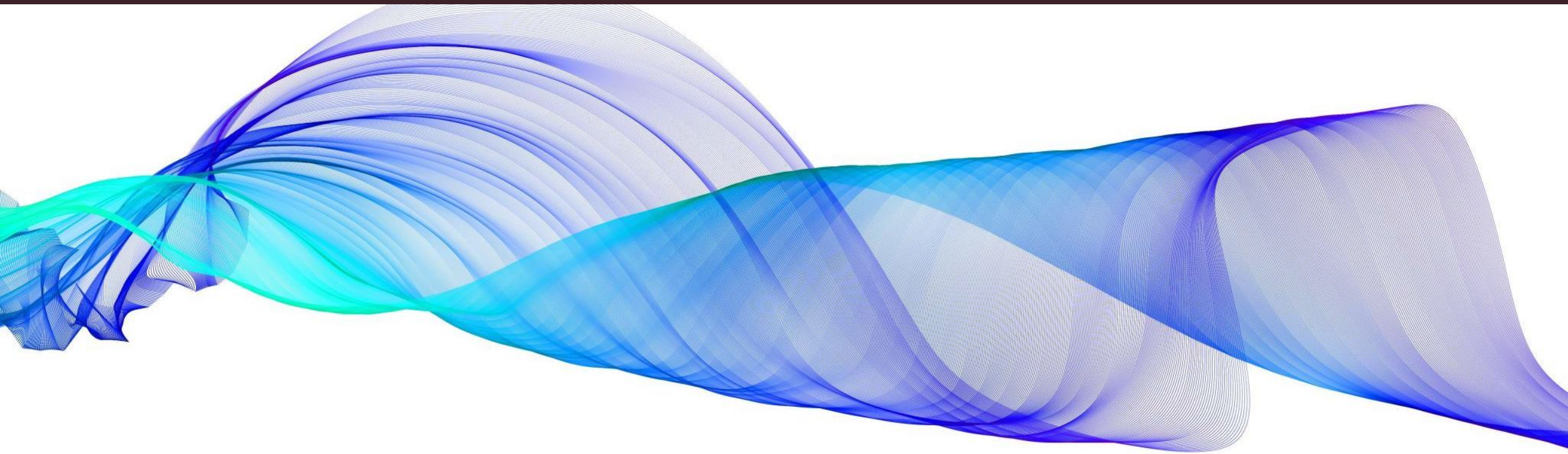
bloom syndrome association

NHS England

Conference presentation

Specialist Genetics clinic for bloom syndrome.

How this clinic has impacted our family



How, when and where.

We were first introduced to the clinic in june 2022.

Our local paediatric consultant had made contact for advice and we were invited to the clinic.

The clinic is at St Thomas hospital in London. Over 200 miles away from our home.

Olivia is seen once a year at the clinic but I have regular contact with the specialist nurses. If she needs to be seen before her Yearly appointment this has also been facilitated.

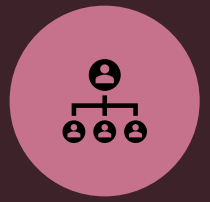
There is also regular contact with the drs and our local consultant.

We also attend check ups in Manchester at the children's hospital with Dr Meyer this is 6 monthly visits.

A day in the clinic

We arrive at clinic and head to the blood room at 830am. The team here are amazing.

We then head up to the rare disease centre where we are met by the team and we are given a list of timings when we are due to see each specialist.



Specialists include Genetics, dentist, dermatologist, dietitian, opthalmology, psychology, occupational therapist, endocrinologist, specialist nurses and there is also now an immunologist joining ththe team.

The waiting room is welcoming and a charity that works with the team ‘amy and friends’ help by bringing games for the children and also are great comfort for the parents.

This is the first time We have sat in a medical room And have been able to talk to families who are on the same journey as us.

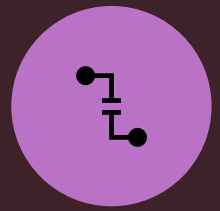
The clinic impact on our family

WE HAVE HAD SO MANY POSITIVE EXPERIENCES SINCE BEING PART OF THIS CLINIC.

WE NOW HAVE A CONSISTENT POINT OF CONTACT THAT ARE SPECIFICALLY INTERESTED IN BLOOM SYNDROME. THIS IS FOR OURSELVES AND FOR THE MEDICS INVOLVED.

WE HAVE STRUGGLED TO HAVE SURVEILLANCE TESTS LOCALLY – THE CLINIC AND TEAM HAVE BEEN ABLE TO ARRANGE THESE ELSEWHERE.

HAVING A PLAN AFTER EACH CLINIC HELPS THE MEDICAL TEAMS LOCALLY.



I DEFINATELY FEEL NOW THERE IS A CONTINUITY OF CARE IN RELATION TO BLOOM SYNDROME.

THE ADVANTAGE OF THE CLINIC IS WE SEE SEVERAL SPECIALISTS IN ONE DAY AND A PLAN IS MADE WHICH SAVES ENDLESS APPOINTMENTS THROUGHOUT THE YEAR.

THE PLAN MADE AT THE MDT IS THEN FORWARDED TO OUR LOCAL CONSULTANT, GP AND OURSELVES SO EVERYONE IS UP TO DATE.

REFERRALS CAN ALSO BE SENT FROM THE CLINIC AND THEY HAVE BEEN GREAT SUPPORT WITH HER EDUCATIONAL NEEDS AND HER MENTAL HEALTH.