

**National Bloom Syndrome Service**

**Referral Form: Bloom Syndrome**

**Rare Diseases Centre**

**First Floor Lift/Stairs B**

**South Wing (Purple Zone)**

Westminster Bridge Road

London, SE1 7EH

CS/TTD Administrator: 0207 188 7188 x 58030

Date of Referral:

Referrer Name:

Referrer Designation:

Referrer Contact Details:

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| **Patient Information**  Surname:  First name:  Date of Birth:  NHS number  Ethnicity:  First Language:  Translator required: **🞏** Y  **🞏** N  Sex: **🞏** M **🞏** F  Diagnosis:  Age of diagnosis:  Genetic Mutation: | Address:  Telephone:  E-mail: | GP:  Local Paediatrician:  Email:  Telephone: | Safeguarding Concerns: Y/N  (please specify if Y)  Social Worker: |

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| --- | --- |
| **Mother:**  Name  D.O.B:  Ethnicity:  **Father:**  Name  D.O.B:  Ethnicity: | **FAMILY TREE** (if possible)  Consanguinity: **🞏** YES (specify on the tree) **🞏** NO |

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| **PRENATAL PERIOD**  Abnormal fetal ultrasound: ⬜ NO ⬜ YES - Please Specify: | |
| **BIRTH**  Term: …………………..  Weight: ……………………………….  Length: ……………………………….  Head circumference: ………….……… | |  |  |  |  |  | | --- | --- | --- | --- | --- | | Congenital cataracts: | ⬜ | NO | ⬜ | YES | | Joint contractures at birth: | ⬜ | NO | ⬜ | YES |   Other (please specify):  Skin: dry or scaly: ⬜ NO ⬜YES |
| **NEUROLOGICAL DISORDERS**  Intellectual disability:  ⬜ None ⬜ Mild ⬜ Moderate ⬜ Severe  Onset: ……………………………………… | Age of walking: …………………………………  Age of loss of walking: …………………………………  **NEUROIMAGING (MRI, CT-scan) :** ⬜ NO ⬜ YES  Result if available: |
| OPHTHALMIC (Eye) DISORDERS  |  |  |  |  |  | | --- | --- | --- | --- | --- | | Pigmentary retinopathy | ⬜ | NO | ⬜ | YES | | Optic atrophy | ⬜ | NO | ⬜ | YES | | |  |  |  |  |  | | --- | --- | --- | --- | --- | | Cataracts | ⬜ | NO | ⬜ | YES |   Other (please specify): …………………………………………... |
| AUDITORY (hearing) ASSESSMENT Sensorineural hearing loss ⬜ None ⬜ Mild ⬜ Moderate ⬜ Severe  Auditory evoked potential ⬜ Not done ⬜ Done  Hearing Aids ⬜ YES ⬜ NO | |
| SKIN EXAMINATION  |  |  |  |  |  | | --- | --- | --- | --- | --- | | Cutaneous photosensitivity | ⬜ | NO | ⬜ | YES | | |
| DENTAL EXAMINATION  |  |  |  |  |  | | --- | --- | --- | --- | --- | | Enamel abnormalities (hypoplasia, cavities) | ⬜ | NO | ⬜ | YES | | Abnormality in shape, in size, in number | ⬜ | NO | ⬜ | YES | | Teeth removed  Local Dentist Name and Contact Details: | ⬜ | NO | ⬜ | YES | | |
| **FEEDING HISTORY**   |  |  |  |  |  |  |  |  |  | | --- | --- | --- | --- | --- | --- | --- | --- | --- | | **Oral** |  | ⬜ | NO | | ⬜ | | YES | | | **NG tube** |  | ⬜ | NO | | ⬜ | | YES | | | **Gastrostomy** |  | ⬜ | NO | | ⬜ | | YES | | |  |  |  | |  | |  | | | |
| |  |  |  |  |  | | --- | --- | --- | --- | --- | | **Known Allergies:** |  | Y/N | Please Specify: |  | |  |  |  |  |  |   **Drug Reaction:** Y/N Please Specify: | |
| |  |  |  |  |  | | --- | --- | --- | --- | --- | | **Skin biopsy:** | ⬜ | NO | ⬜ | YES | |  |  |  |  |  | | **Genetic test:** | ⬜ | NO | ⬜ | YES | | **Available Results:**  **DNA Results:**  **Chromosome Breakage Results:** |  |  |  |  | | |

**Malignancies/Haematology**

**Screening Programme in Place:** ⬜ NO ⬜ YES **Programme Details:**

**Family History of Cancer:**

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**History of Previous Malignancies:** ⬜ NO ⬜ YES **Details:**

**Haematologist Details:**

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**Endocrinology/Immunology**

**Diabetic:**

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| ⬜ | NO | ⬜ | YES |

**Recurrent Infections: Immunotherapy:** ⬜ NO ⬜ YES

|  |  |  |  |
| --- | --- | --- | --- |
| ⬜ | NO | ⬜ | YES |

**Please complete referral form fully and e mail to** [**gst-tr.cs-ttd@nhs.net**](mailto:gst-tr.cs-ttd@nhs.net) **along with any genetic testing results and clinical letters that are available.**

**Any queries: please contact Dr Shehla Mohammed – National Lead for Bloom Syndrome or Paula Sullivan/Phillipa Sellar – Nurse practitioners for Bloom Syndrome on** [**gst-tr.cs-ttd@nhs.net**](mailto:gst-tr.cs-ttd@nhs.net)