

**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:

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

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

|  |
| --- |
| **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 ⬜ SevereOnset: ……………………………………… | 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 |

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| Cataracts | ⬜ | NO | ⬜ | YES |

Other (please specify): …………………………………………... |
| AUDITORY (hearing) ASSESSMENTSensorineural hearing loss ⬜ None ⬜ Mild ⬜ Moderate ⬜ SevereAuditory evoked potential ⬜ Not done ⬜ DoneHearing Aids ⬜ YES ⬜ NO |
| SKIN EXAMINATION

|  |  |  |  |  |
| --- | --- | --- | --- | --- |
| Cutaneous photosensitivity | ⬜ | NO | ⬜ | YES |

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| DENTAL EXAMINATION

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| --- | --- | --- | --- | --- |
| Enamel abnormalities (hypoplasia, cavities) | ⬜ | NO | ⬜ | YES |
| Abnormality in shape, in size, in number | ⬜ | NO | ⬜ | YES |
| Teeth removedLocal Dentist Name and Contact Details: | ⬜ | NO | ⬜ | YES |

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| **FEEDING HISTORY**

|  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- |
| **Oral** |  | ⬜ | NO | ⬜ | YES |
| **NG tube** |  | ⬜ | NO | ⬜ | YES |
| **Gastrostomy** |  | ⬜ | NO | ⬜ | YES |
|  |  |  |  |  |

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| --- | --- | --- | --- | --- |
| **Known Allergies:**  |  | Y/N | Please Specify: |  |
|  |  |  |  |  |

 **Drug Reaction:** Y/N Please Specify: |
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| --- | --- | --- | --- | --- |
| **Skin biopsy:** | ⬜ | NO | ⬜ | YES |
|  |  |  |  |  |
| **Genetic test:** | ⬜ | NO | ⬜ | YES |
| **Available Results:** **DNA Results:****Chromosome Breakage Results:** |  |  |  |  |

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**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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| --- | --- | --- | --- |
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**Endocrinology/Immunology**

**Diabetic:**

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

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

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

**Please complete referral form fully and e mail to** **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**