

PREDICT Study FORM02: Phenotype

Ethics approval: SCHN 2020/ETH00634

Contact: -----

Study admin use only

REDCap ID - **General**1) Date of exam DD / MM / YY**Patient information**

If available, place patient sticker over dashed outline.

PLACE PATIENT
STICKER HERE2) First name 3) Last name 4) Other names 5) Date of birth DD / MM / YY**Examiner information**6) Name

7) Position

 JMO Fellow Consultant Other: **Anthropometrics**8) Height cm9) Weight kg10) Head circumference cm**Asymmetry**

11) Asymmetry

 None Head Face Limbs**Cranium**

12) Scalp tumours

 Yes No

13) Hair type

Mark all that apply

 Normal Brittle Sparse/fine Coarse

14) Frontal bossing (prominent forehead)

 Yes No**Eyes**

15) Cataract

 Not examined Yes No

16) Visible nerve fibers on cornea

 Not examined Yes No

17) Photosensitivity

Mark all that apply

 No Yes, eyes Yes, skin

18) Eye spacing

 Normal Orbital hypotelorism Orbital hypertelorism

19) Eye size

 Normal Microphthalmia Macrophthalmia

20) Palpebral fissure slant

 Normal Downslanted Upslanted

21) Epicanthal folds

 Yes No

22) Ptosis (drooping eyelid)

 Yes No

23) Abnormalities of eye control

Not explained by primary tumour. Check all that apply.

 None Strabismus Nystagmus Amblyopia Other:

24) Vision loss

Not explained by primary tumour. Check all that apply.

 None Colour vision deficiency Partial vision loss (bilateral) Hemianopsia Complete blindness

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REDCap ID - **Ears**

25) Ear lobule creases or pits

- Yes
 No

26) Posterior helical ear pits

- Yes
 No

27) Abnormal ear shape

If yes, please describe

- Yes:
 No

Oral region

28) Jaw position

Mark all that apply

- Normal
 Micrognathia
 Retrognathia
 Prognathia

29) Lips

Mark all that apply

- Normal
 Periorificial papilloma
 Perioral hyperpigmentation
 Mucosal neuromata

30) Oral cavity

Mark all that apply

- Normal
 Short superior frenulum
 Cleft palate
 Hard palate high/short/narrow

31) Oral mucosa

Mark all that apply. If other, please comment.

- Normal
 Leukoplakia
 Gingival overgrowth
 Papilloma / neuroma
 Papular fibromatosis
 Cobblestone
 Other:

32) Tongue

Mark all that apply

- Normal
 Macroglossia
 Geographic
 Lobulated
 Glossoptosis
 Ankyloglossia

Oral region continued on right

Oral region (continued)

33) Oral pigmentation

Mark all that apply. If other, please comment.

- Normal
 Mucocutaneous pigmentation
 Other:

34) Dental abnormalities

Mark all that apply. If other, please comment.

- Normal
 Hypodontia (congenital)
 Hyperdontia
 Unerupted teeth
 Impacted teeth
 Enamel pits
 Other:

35) Has a mouth X-ray been performed?

Mark all that apply. If other, please comment.

- No, or not known
 Yes, with no abnormalities identified
 Yes, with odontogenic keratocysts detected.
 Yes, with other abnormalities detected:

Thorax

36) Supernumerary or widely-spaced nipples

- Yes
 No

37) Gynaecomastia (in males)

- Yes
 No
 Not applicable (patient is female)

Abdomen

38) Umbilical hernia

- Yes
 No

Joints and skeleton

39) Neck

Mark all that apply

- Normal
 Short neck
 Pterygium colli
 Redundant neck folds

Joints and skeleton continued on next page

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REDCap ID - **Joints and skeleton** (continued)

40) Chest wall deformities

Mark all that apply. If other, please comment.

- None
- Pectus excavatum
- Pectus carinatum
- Cleft sternum
- Other:

41) Scoliosis of spine

- Yes
- No

42) Limbs

Mark all that apply

- Normal
- Reduced range of movement
- Hypermobile joints
- Absent radius
- Other limb abnormalities:

Hands and feet

43) Digit abnormalities

Mark all that apply

- None
- Polydactyly
- Syndactyly
- Clinodactyly
- Camptodactyly
- Other:

44) Palm / plantar

Mark all that apply

- Normal
- Palmar/plantar pits
- Single palmar crease
- Single plantar crease

45) Nails

Mark all that apply

- Normal
- Hypoplastic
- Nail pits

Genitalia

46) Precocious puberty

- Not applicable (prepubertal)
- No: puberty is age-appropriate
- Yes: precocious puberty present
- Not examined

Genitalia continued on right

Genitalia (continued)

47) Abnormal genital pigmentation

- Yes
- No
- Not examined

48) Ambiguous genitalia

- Yes
- No
- Not examined

49) Undescended testicle (if patient is male)

- Yes
- No
- Not examined

Skin

50) Telangiectasia

- Yes
- No

51) Tumours

Mark all that apply

- None Neurofibromas
- Lipomata Schwannomas
- Pilomatrixomas
- Sebaceous Adenomas
- Other:

52) Blue naevus

- Yes
- No

53) Pigmentation

Mark all that apply. If other, please comment.

- Normal
- Café-au-lait spots
- Axillary freckling
- Hyperpigmentation
- Hypopigmentation
- Inguinal freckling
- Other:

If Café-au-lait spots, please comment on number and size

54) Palmoplantar hyperkeratosis

- Yes
- No

55) Thin skin, or generalised skin atrophy

- Yes
- No

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REDCap ID - **Neurological signs**

(Not explained by primary tumour)

56) Ataxia

 Yes No

57) Cranial nerve palsies

 Yes No**Endocrine**

58) Enlarged thyroid

 Yes No

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