COST Imprinting Disorder questionnaire and generic examination tool: Health professional to patient version 02022015

Participant's full name: ________________________________

Imprinting disorder (clinical diagnosis) ________________

Confirmed with genetic result Yes ☐ No ☐

If yes specify ________________________________

Laboratory that made diagnosis ____________________________

Participant date of birth d d ☐☐ m m ☐☐ y y ☐☐

Sex M=Male F=Female ☐

Address and post/ZIP code ☐

Country of birth ☐

Ethnic origin ☐

HISTORY AND EXAMINATION

Visit date d d ☐☐ m m ☐☐ y y ☐☐

Hospital number...

COST ID number...

present at interview: mother ◻ father ◻ patient/proband ◻

NAME of DOCTOR completing the form

ADDRESS

EMAIL

Consent for sharing of data Yes ☐ No ☐

Date

Health Professional taking consent

Consent for PHOTO (head and neck (full and profile), hands/feet/full body shots

Yes ☐ No ☐

Date

Health Professional taking consent

Section 1

Conception

At time when patient was conceived Gravida Para

1. Were there any problems conceiving you/the proband or other children (i.e. child's siblings)?

History of Infertility ☐Yes ☐No ☐Unknown
Comments:

If Yes

Was 'Assisted Reproductive Technology' (ART) used in your/the proband's pregnancy?
☐Yes ☐No ☐Unknown

If yes, what was used?

If no to ART, was other medication used? ☐Yes ☐No ☐Unknown

2. Was there certainty about your/the proband's date of conception?
☐Yes ☐No ☐Unknown

Comments:

3. Are you/the proband a monozygous twin? ☐Yes ☐No ☐Unknown

4. Are you/the proband a dizygous twin? ☐Yes ☐No ☐Unknown

Pregnancy of the proband

5. Was growth less than expected during pregnancy?
(i.e. intrauterine growth retardation diagnosed in pregnancy) ☐Yes ☐No ☐Unknown

If yes

provide approximate stage in the pregnancy when first noted;
1sr trimester 2nd trimester 3rd trimester

6. Was growth more than expected during the pregnancy?
(i.e. excess growth diagnosed in pregnancy) ☐Yes ☐No ☐Unknown

If yes

provide approximate stage in the pregnancy when it was first noted
1st trimester 2nd trimester 3rd trimester

7. Tick any of the following noted during pregnancy

Bleeding
Polyhydramnios
Oligohydramnios

Comments

Severe nausea and vomiting
High BP of mother
Known infections
Congenital anomalies identified in pregnancy

8. Did you/your mother drink excess alcohol (more than 4 units per week) during the pregnancy?

Excess Alcohol ☐Yes ☐No ☐Unknown

9. Folic acid taken in pregnancy ☐Yes ☐No ☐Unknown

Dose if known

Provide approximate stage in the pregnancy when it was started and continued

before conception 1st trimester 2nd trimester 3rd trimester

10. Other medication, smoking history or other toxic drugs in pregnancy
(including IVF drugs if applicable)

Drugs used / smoked a day

11. Occupational exposure of mother and father (record employment)

Exposure comments

Delivery of the proband

12. Type of Delivery (Tick)

Spontaneous
C-section (indicate if emergency or elective)
Forceps
Other

13.

Duration of gestation of pregnancy weeks
State how you calculate this i.e. weeks from amenorrhoea OR from implantation

14. Placenta (after-birth):

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Placenta; visual inspection:	☐Normal	☐Abnormal	☐Unknown
Placenta described as big for gestation	☐Yes	☐No	☐Unknown
or small for gestation	☐Yes	☐No	☐Unknown

Placenta weight in gms if known ……………………………………………………………………………………………………

Comments

At birth of proband

15. Growth parametres at birth (if recorded):

Birth weight in (gms)	………………gms	………….SDS
Birth length (cm)	………………cm	…………..SDS
Birth head circumference (cm)	……………..cm	………….SDS

16. Apgar score at 1st min . …….., 5th min. ……….., 10th min. ………….

17. Asymmetry of growth of the body (if noted at brith) ☐Yes ☐No ☐Unknown
Describe

18. Any unusual features noted or
congenital anomalies diagnosed at birth ☐Yes ☐No ☐Unknown

If yes describe

comments

19. Direct questions Did you/the proband have a history of :

Blood sugar issues?

- episodes of hypoglycaemia	☐Yes	☐No	☐Unknown
- episodes of excessive sweating	☐Yes	☐No	☐Unknown
- episodes of hyperglycaemia	☐Yes	☐No	☐Unknown
Exomphalos	☐Yes	☐No	☐Unknown
Umbilical hernia	☐Yes	☐No	☐Unknown
Diastasis Recti	☐Yes	☐No	☐Unknown
Macroglossia	☐Yes	☐No	☐Unknown
Small tongue	☐Yes	☐No	☐Unknown
Micrognathia	☐Yes	☐No	☐Unknown
Cleft palate	☐Yes	☐No	☐Unknown
High arched palate	☐Yes	☐No	☐Unknown
Prominent jaw	☐Yes	☐No	☐Unknown
Hypogonadism	☐Yes	☐No	☐Unknown
Hypospadias repair (males)	☐Yes	☐No	☐Unknown
Undescended testes in male	☐Yes	☐No	☐Unknown
Vaginal anomalies	☐Yes	☐No	☐Unknown
Other genital anomalies	☐Yes	☐No	☐Unknown

Describe:

Hypotonia	☐Yes	☐No	☐Unknown
Stridor	☐Yes	☐No	☐Unknown
Scoliosis	☐Yes	☐No	☐Unknown
Joint hyper-extensibility	☐Yes	☐No	☐Unknown
Incurved of little fingers	☐Yes	☐No	☐Unknown
Joint contractures	☐Yes	☐No	☐Unknown
Macrodigits	☐Yes	☐No	☐Unknown
Heart defects	☐Yes	☐No	☐Unknown
Renal defects	☐Yes	☐No	☐Unknown
Congenital anomalies unspecified*	☐Yes	☐No	☐Unknown
Abnormal brain scan**	☐Yes	☐No	☐Unknown
Delayed closure of anterior fontanel (age) ***	☐Yes	☐No	☐Unknown
Craniosynostosis ***	☐Yes	☐No	☐Unknown
Dislocated elbow	☐Yes	☐No	☐Unknown

Comments or other features (*) (**) (***)

Development of proband

20. Were there worries about early development?	☐Yes	☐No	☐Unknown
21. Were there worries about reaching normal motor milestones	☐Yes	☐No	☐Unknown
22. Were there any concerns about speech development	☐Yes	☐No	☐Unknown

23. In more detail

Age in months when

Sat unaided……………………. ☐Unknown

Crawled……………………….. ☐Unknown

Walked unaided……………….. ☐Unknown

First said a few words………….. ☐Unknown

Visual difficulties	☐Yes	☐No	☐Unknown
Hearing difficulties	☐Yes	☐No	☐Unknown

If yes provide any details

comments

Schooling

24. Special educational needs at school

☐Yes ☐No ☐Unknown

25. What type of school did you/the proband attend?

comments

26. In more detail

Nursery:	main stream ☐		special needs nursery ☐
Primary School:	main stream ☐	mainstream with special help ☐	special needs education ☐
Secondary school:	main stream ☐	main stream with special help ☐	special needs education ☐

Indicate nature of special help……………………………………………………………

Statement of educational needs? ☐Yes ☐No ☐Unknown

Age of leaving education in years (if appropriate) ………………………………

27. Do you/the proband have any national education qualifications (country specific)?

Other educational qualifications:……………………………………………………………………………………………………

28. Behaviour

Do you consider your child/you as proband to have any unusual behaviours?
☐Yes ☐No ☐Unknown

With the sleep pattern?	☐Yes	☐No	☐Unknown
With the anger control?	☐Yes	☐No	☐Unknown
Ability to play with friends?	☐Yes	☐No	☐Unknown

Other specify

Family History

Pedigree to third generation relatives with date of births, first names, approximate height and weight, birth weight and lengths (if known) and significant illnesses warranting an appointment in hospital. Ask about periods of prolonged starvation and workplace for parents and grandparents. Indicate who gave the information.

Medical History of the proband

Feeding history

Neonatal phase

29.

Feeding difficulties as a neonate ☐Yes ☐No ☐Unknown

Poor suckling ☐Yes ☐No ☐Unknown

Normal eye gaze when feeding ☐Yes ☐No ☐Unknown

Crying for food ☐Yes ☐No ☐Unknown

Feeding via a tube ☐Yes ☐No ☐Unknown

If yes to a tube, provide information including age when it improved

30. Feeding via a gastrostomy? ☐Yes ☐No ☐Unknown

If yes provide information including age when it was reversed

Name medication

Appetite history after 2 years

31. Poor appetite (no crying for food) ☐Yes ☐No ☐Unknown

If yes provide information and ages when it improved (if it did)

32. Excessive appetite at any time (including excessive crying for food or 'stealing' food compared to other family members)

☐Yes ☐No ☐Unknown

If yes provide information and ages when it improved (if it did)

33. Gastroesophageal reflux/ heartburn ☐Yes ☐No ☐Unknown

If yes provide information such as age and if it improved

34. Was reflux confirmed with PH probe ☐Yes ☐No ☐Unknown

35. Have you/your child had/have received 'oral desensitisation' therapy

☐Yes ☐No ☐Unknown

36. Severe constipation warranting referral to a doctor ☐Yes ☐No ☐Unknown

If yes provide information such as age and if it improved

37. Drugs used for appetite ☐Yes ☐No ☐Unknown

38. Drugs used to stimulate/promote appetite ☐Yes ☐No ☐Unknown

General medical history in childhood, teenage years and adulthood

39. Any medical conditions diagnosed:

List conditions requiring attendance at hospital

1.
2.

3.

comments

40. Growth

List any known **height, head and weight measurements** at different ages recorded (proband) ideally at ages specified but change depending on data/ age of proband

	Height cm	Weight kg	Head cm	Date taken
Earliest after birth				
Age 1				
Age 2				
Age 5				
Age 8				
Age 10				
Age 15				
Age 20				

Age when stopped growing (height) ....

41. Puberty

Girls

At what age was pubic hair was noticed ....

At what age was breast development noticed ....

At what age did regular periods started ....

Assessment of age of puberty (female)

Boys,

At what age puberty started ....

At what age pubic hair was noticed ....

At what age growth was complete ....

Was puberty at the same time as peers at school? ....

Assessment of boy's age of puberty

Medication related to puberty ☐Yes ☐No ☐Unknown

details

Drug History (excluding growth hormone)

42. Medication for significant medical issues

List including ages when taken if known (see below for GH medication)

comments

Growth Hormone therapy (GH)

43. Did you/the proband receive Growth Hormone Therapy: ☐Yes ☐No ☐Unknown

List age when started.....................
Age when stopped..............................
Dosages – where possible with ages

44. Any other issues relating to GH?

comments

45. Was ever IGF1 measured? ☐Yes ☐No ☐Unknown

details

Examination checklist Generic for all imprinting disorders; Date of examination……………………………

1. Head circumference in cm (+ SDS?)

SDS

2. Height in cm (+ SDS?)

3. Sitting height in cm (+SDS)

4. Weight (preferably in light clothing such as shorts & t-shirt) (+ SDS?)

5. BMI (+SDS) ……………………………………

6. Waist circumference (+SDS)

PUBERTAL ASSESSMENT only relevant for adolescents (Tanner stages 1 to 5)

The pubertal assessment will be completed by the doctor. For any person preferring not to be examined a self-complete questionnaire will be provided.

Boys

Testicular volume (Orchidometer)

Genital stage (Tanner stage)

Pubic hair (Tanner stage)

Penis size

Girls

Breast development (Tanner stage)

Pubic hair (Tanner stage)

Morphology exam- - *take photos of face in exam (try and ascertain childhood photos)*

Face Overall gestalt ☐Yes ☐No ☐Unknown

Name syndrome ……………………………………………………………………………

Dysmorphic features list

Familial ………………

Non-familial………………………

Triangular facies? ☐Yes ☐No ☐Unknown

Prominent jaw ☐Yes ☐No ☐Unknown

Prominence of forehead at 2 years (by photo if necessary) ☐Yes ☐No ☐Unknown

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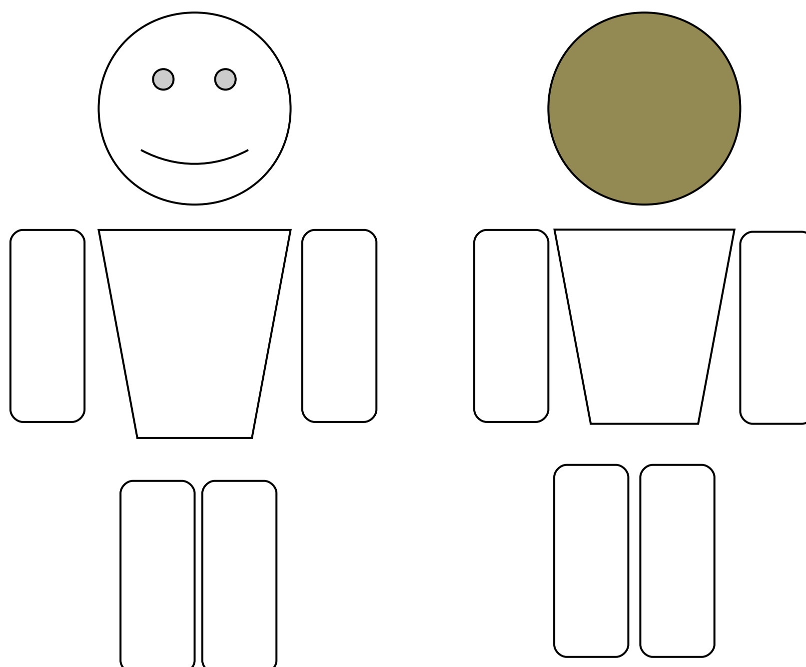
Skin Normal: ☐Yes ☐No ☐Unknown

Describe birth marks, neurocutaneous stigmata or cafe au lait spots
Pigmentary anomalies
Cutis laxa, tight skin, or unusual texture
Acanthosis nigricans
Plaques
Dimples
etc

Front **Back**



Body asymmetry on examination (more than just the face; requires size difference of >1 cm:

☐Yes ☐No ☐Unknown

Larger side: ☐Right ☐Left ☐Unknown

Measure asymmetry where possible and describe

Cranium shape normal ☐Yes ☐No ☐Unknown

Describe skull shape if abnormal e.g. brachycephaly

Ears Normal shape ☐Yes ☐No

Comments

Eyes Any unusual features noted

Nose Any unusual features noted

Philtrum Describe, e.g. long/short/smooth

Mouth

Tongue, normal size ☐Yes ☐No ☐Unknown

Comment e.g. asymmetry:

Palate, normal ☐Yes ☐No

Gums normal ☐Yes ☐No

Teeth normal ☐Yes ☐No

Chin Normal ☐Yes ☐No

If no is there micrognathia? ☐Yes ☐No

If no is there macrognathia? ☐Yes ☐No

Hands and feet

Size of hands (measure from wrist crease to tip of middle digit L and R ☐

Measure from proximal middle finger crease to tip L and R ☐

Hands

Normal examination ☐Yes ☐No

If no

5th finger clinodactyly	☐Yes	☐No
Finger contractures	☐Yes	☐No
Digitalisation of thumb	☐Yes	☐No
Missing digits	☐Yes	☐No
Macrodigit	☐Yes	☐No
Syndactyly	☐Yes	☐No
Nails, normal	☐Yes	☐No

comments

Feet

Normal examination ☐Yes ☐No

If no

Prominence of heels	☐Yes	☐No
Syndactyly	☐Yes	☐No
Sandal gap present	☐Yes	☐No

comments

Chest Normal examination ☐Yes ☐No

Comments

Heart Normal examination ☐Yes ☐No

Comments

BP: Reading

Abdomen Normal examination ☐Yes ☐No

Comments

Back Normal examination ☐Yes ☐No

Comments

Nervous System Normal examination ☐Yes ☐No

If no

Hypotonia	☐Yes	☐No
Muscle strength	☐Yes	☐No
Muscle mass	☐Yes	☐No
Reflexes	☐Yes	☐No

Comments

Congenital anomaly identified ☐Yes ☐No

If yes indicate number of congenital anomalies by circling the number 1 2 3 4 5

comments

Communication within normal spectrum ☐Yes ☐No

Indicate type of problem if no

Speech delay/limited	Mild	Moderate	Severe
Hearing deficit	Mild	Moderate	Severe
Visual problem	Mild	Moderate	Severe
Understanding	Mild	Moderate	Severe

comments

Intellectual abilities within normal spectrum ☐Yes ☐No

If no Indicate type of development problem

Global delay Mild Moderate Severe

Space for other comments re examination

Investigation consider **including date taken**

1. Bone age
2. Bone density
3. Chromosome breakage
4. Biochemistry
5. Fat metabolism
6. Hormone assessment
7. Skeletal survey
8. Liver function tests
9. Calcium metabolism
10. Growth Hormone
11. Glucose metabolism

WELLBEING (questions) - I did not change this part, as it refers rather to elder patients

Health

1. Would you say that your health is excellent, good, fair, or poor?
   1. Excellent
   2. Good
   3. Fair
   4. Poor

☐

School (if currently at school)/Job

2. How do you feel about school/job at present?
   1. I like it a lot
   2. I like it a bit
   3. I don't like it very much
   4. I don't like it at all

☐

Out of place

3. Here are some statements about your life. Please tell me if you agree or disagree with them:
   1. Strongly agree
   2. Agree
   3. Disagree
   4. Strongly disagree

I feel like an outsider or left out of things ☐

I feel awkward and out of place ☐

I feel lonely ☐

Life satisfaction

4. Here is a picture of a ladder. The top of the ladder,10, is the best possible life for you and the bottom, 0, is the worst possible life for you. In general, where on the ladder do you feel you stand at the moment? Please point to the number that best describes where you stand.'

☐☐

10
9
8
7
6
5
4
3
2
1
0