CHARGE Syndrome Clinical Database Project (CSCDP) Section Descriptions

Note: At the end of each section, you can choose to Submit or Save and Return Later

Save and Return Later: choose this if you are not finished with the section or if you do not want to go on to the next section at this time:

You will be asked to provide an email address for the return link to be sent to

You will be given a Return Code: Write it down – it will not be in the email you receive

When you are ready to return, use the return link in the email and enter your Return Code

You will be returned to the top of the section you Saved. Previously entered data will be there and can be edited. Complete the section (or scroll to the end of the section if it was complete) and Submit.

Submit: this will submit the entered data and take you to the top of the next section. Once a section is submitted, you will not be able to return to it. (If you are not finished, Save and Return Later). Sections must be completed in order. If you are done with a section but do not want to complete the next one, you may Submit the section and then scroll down and Save and Return Later in the following section.

CSCDP SECTIONS

Recruitment/Consent and HIPAA You will be asked

- Name and birthdate of person with CHARGE
- Your name
- Your relationship to person with CHARGE
- To read the consent form and HIPAA (privacy) information. You must Accept both forms to participate in the project

Time to read and complete: about 10 minutes

Demographics. You will be asked

- Name and birthdate of the person with CHARGE
- Name and contact information for person completing the surveys
- Birthdates of biological mother and father
- Whether anyone else in the extended family has CHARGE or features of CHARGE syndrome

Time to complete: about 5 minutes

Birth History of person with CHARGE. You will be asked

- Place of birth (city, state, hospital)
- Birth date, weight, length, & head circumference
- Prenatal history
- Any significant ultrasound findings
o Prenatal testing, testing outcomes
o Age at diagnosis, certainty of diagnosis
o If deceased, age and cause of death

Time to complete: 10-15 minutes.

**Major Diagnostic Characteristics.** You will be asked about

- Coloboma – location in each eye
- Choanal atresia/stenosis
- Cranial nerves
  - Smell
  - Eye movement
  - Tongue movement
  - Facial palsy
  - Feeding problems
  - Shoulder shrugging
- Ears
  - External ear shape
  - Middle ear - bones, infections
  - Inner ears -cochlea, semicircular canals
  - MRI and CT findings

You will have an opportunity to **upload photos** of eyes (R, L & both), face (showing facial palsy) and ears (R & L). Any photos you choose to upload may be used for publications or teaching.

Note: vision, hearing and surgeries are covered in other sections

Time to complete: 20-30 minutes.

**Minor Characteristics & Other Findings.** You will be asked about

- Cleft lip and palate
- Teeth
- Heart
- Trachea and esophagus
- Kidneys
- Genitalia
- Hypotonia
- Hockey stick palmar crease
- Craniosynostosis

Note: surgeries are covered in separate section.

Time to complete: 10 minutes

**Skeletal Features** You will be asked about

- Skull, including craniosynostosis
- Hands & fingers
- Feet & toes
- Arms & Legs
- Neck & shoulders
- Back & ribs
- Joints

Time to complete: 5-10 minutes

**Genetic testing.** You will be asked about tests and results for
- CHD7 - when, what lab, specific results
- Chromosomes
- FISH for del22q
- Other genetic testing (microarray, CMA, WES)

You will have an opportunity to **upload scanned copies** of test results.

Time to complete 5-15 minutes.

**Surgeries and Hospitalizations.** You will be asked
- Total number of months your child spent in the hospital in the first three years of life
- All of your child’s surgeries.
- **For each type of surgery** (choanal atresia, heart, PE tubes, etc.) you will be asked
  - age at first surgery
  - age at second surgery
  - total number of each type of surgeries
  - age at most recent surgery

Time to complete: 15-30 minutes depending on how many surgeries and if you have all of the information handy.

**Vision.** You will be asked about
- Loss of visual field (blind spots)
- Visual acuity (e.g. 20/200) for each eye
- Whether the person with CS wears glasses (spectacles) and what the glasses correct (near or farsightedness, etc.).

Time to complete: 5 minutes

**Hearing.** You will be asked
- Degree of hearing loss
- Type of hearing loss (conductive, sensorineural)
- Speech reception threshold (unaided and aided)
- What appliances are used (aids, implants)
- Whether appliances help
- Auditory processing disorder

You will have the opportunity to upload scanned audiograms for your child. We would like:
  - Earliest/best early testing
  - Audiograms which show any apparent changes in hearing
  - Most recent audiogram

Time to complete: 10 minutes plus upload time

Milestones and Growth. You will be asked
- Motor milestones – age at which your child could
  - Sit, crawl, cruise, walk, run, etc.
- Toilet training – age of training for urine and feces, day and night
- Language – age of first word and/or sign, two words and sentences
- Growth over time – you can enter up to 40 measurements
- Height, weight and head circumference

You will have the opportunity to upload scanned copies of your child's growth charts.
- Growth hormone and puberty hormone
  - Was testing done
  - Were hormone given, for how long

Time to complete: 30 minutes or more.

Neurology. You will be asked
- Seizures
- Headaches
- Migraines
- Abdominal migraines
- Brain imaging (MRI or CT)
  - Age of testing
  - Findings
    - Brain
    - Inner ear, including nerves, cochlea and semicircular canals
- Body temperature regulations

Note: We are not asking about behavior at this time

Time to complete: 5-10 minutes

Medications, Bone Health and Sleep. You will be asked
- Checklist of features and conditions (behavior, heart, reflux, etc) that require medications to treat
  - At this time, we are not asking for any medication specifics
- The total number of medications your child is currently taking
- If bone age or DEXA scans have been done. If so, when and results
- If your child has scoliosis, at what age, and how it has been treated
Sleep issues & treatments – check off which issues your child has
Time to complete: 20 minutes

Photos and Comments
- Does the individual with CS have any additional birth defects or features we did not include
- You will have the opportunity to upload up to 10 additional photos of the individual with CS. For example at different ages or showing specific features or behaviors.
- You will be asked to include a description of each photo

Note: any photos you choose to upload may be used for medical publications or teaching
- You will be invited to comment on the survey – including suggestions for other categories or issues you would like us to address in the future
Time to complete: 5-30 minutes depending on photos and comments