

CoRDS Registry

Coordination of Rare Diseases
at Sanford



Coordination of Rare Diseases at Sanford (CoRDS):

An international patient registry at Sanford Research

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CdLS Foundation Webinar

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About CoRDS

Mission: To accelerate research into rare diseases

Goal: To establish an international rare disease patient registry for all rare diseases

- Create resource of contact information and clinical data on individuals diagnosed with any rare disease to enable a comparative analysis across diseases
- Provide a mechanism by which researchers can identify and contact patients interested in participating in research

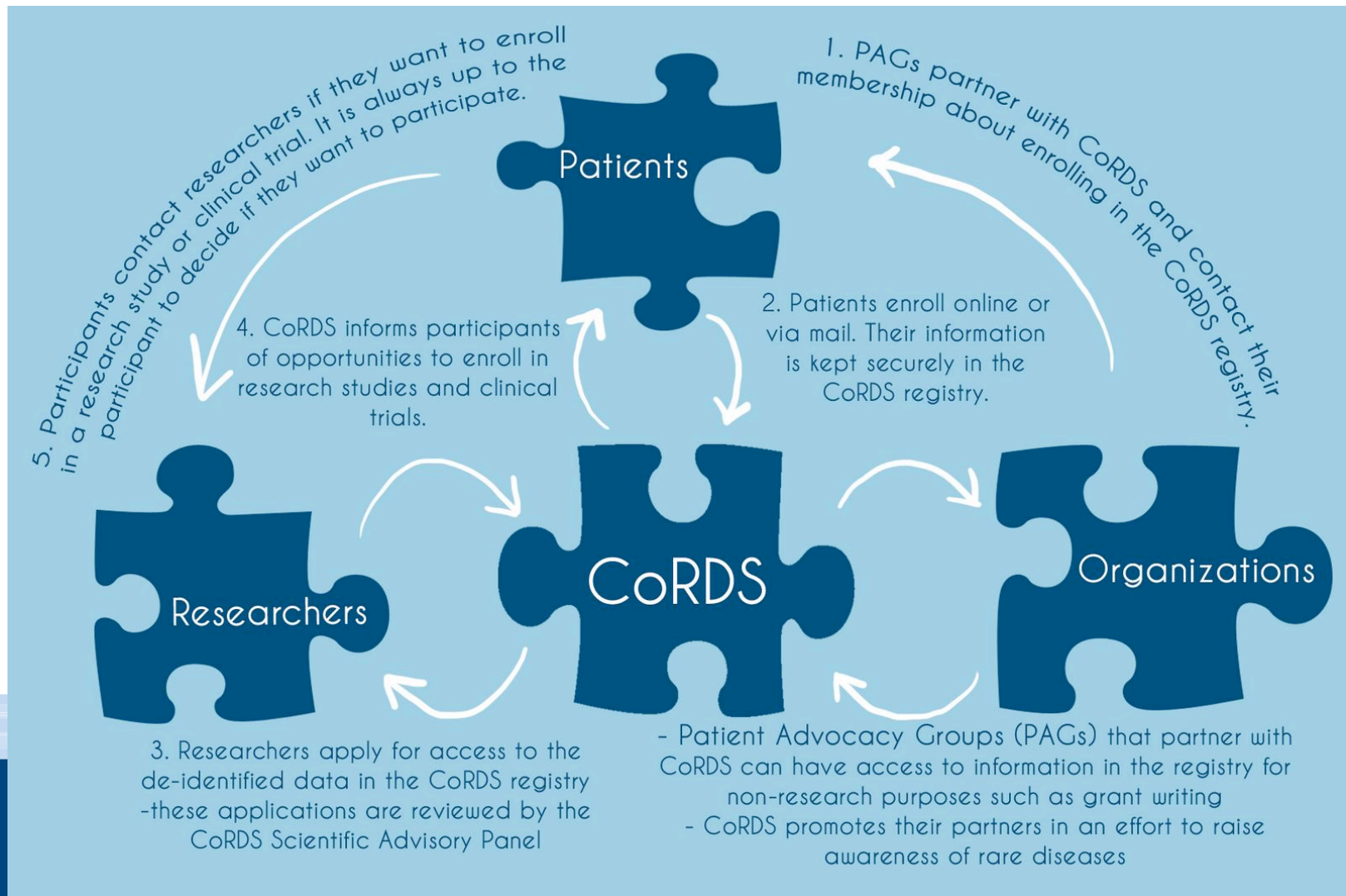
*The term “disease” is used to encompass all rare conditions.

What makes CoRDS unique?

- First of its kind; central resource for all rare diseases; allows for comparative analysis across diseases
- Unique collaboration with patient groups to establish a registry and customize a questionnaire
- Any researcher with IRB approval would have access to the information pending review from advisory panel
- Enrollment option for individuals who are not represented by a rare disease organization and those who are undiagnosed
- **No cost** for patients and families to enroll, no cost for researchers to access, no cost for patient groups to start a registry

Who is involved?

Patients, Researchers & Patient Groups



How are patients & families involved?

- Participants that enroll can contribute information to the CoRDS database by filling out a brief questionnaire
- Participants may learn about research opportunities for which they are eligible
- When notified about research opportunities, they have the ability to decide whether or not they would like to participate

Why would an individual want to participant in CoRDS?

- Provides individuals an opportunity to be informed of research studies and clinical trials for which they are eligible
- Provides researchers with a central resource for the identification and more rapid recruitment of potential research participants
- Has the potential to accelerate research into rare diseases
- No financial cost, not difficult or time consuming to enroll



What is involved after enrollment?

- CoRDS requests and reminds participants to complete their enrollment & update their information annually.
- There is no cost to enroll
- No obligation to participate in research - participants are free to decline any or all research opportunities
- Participants can change preferences and withdraw at any time.

How do researchers access CoRDS?

- Submit brief application to CoRDS Scientific Advisory Board
- Login and access de-identified data
- Identify participants that are eligible for specific research studies
- CoRDS personnel contact participants on behalf of the researcher



How are Patient Groups involved?

- Organizations without a registry can partner to catalog important data
- Flexible to accommodate needs of stakeholder ie customized questionnaires, access to data for non-research purposes, etc.
- No cost for organization to establish a registry
- IRB and IT personnel in-house, software infrastructure for secure data collection & management

CoRDS Patient Registry for the Cornelia de Lange Syndrome Foundation

- Launched August 2014
- Disease-specific registry for individuals diagnosed with Cornelia de Lange Syndrome
- Utilizes common data elements, as well as disease-specific data elements
- Cornelia de Lange Syndrome Foundation has access for non-research purposes and is involved with researcher review process

CoRDS-CdLS Registry Metrics

- Currently the Cornelia de Lange Syndrome Foundation's disease-specific registry includes:
 - 41 fully enrolled participants
 - 44 participants in screening
- Participants have enrolled from
 - 22 US States
 - 2 Countries (United States and India)



Enrollment Options



1. Online Enrollment

- Web-based enrollment platform (April 2012)

2. Mail-Based Enrollment

3. In-person Enrollment

- Patient Advocacy Group annual meetings
- Sanford Health clinics

How do I enroll?

Step 1:

- Participant completes CoRDS Registry Screening form on CoRDS website: sanfordresearch.org/cords
- Participant receives unique username & password via email or receives their forms by mail



CoRDS Registry Form

If you are interested in enrolling in the Cords Registry, please complete the brief screening form below and click submit. You may indicate below if you'd like to enroll online or by mail.

After submitting this form, CoRDS personnel will send you the information you need to complete to enroll if you would like to enroll online, cords personnel will send you an email with a username and password needed to log-in and enroll on the secure site. If you would like to enroll by mail, cords personnel will send you the forms necessary for you to enroll.

Please submit the following:

*Participant First Name:	<input type="text"/>
*Participant Last Name:	<input type="text"/>
*Participant Date of Birth(MM/DD/YYYY):	<input type="text"/>
*Diagnosis:	<input type="text"/>
Parent or Legally Authorized Representative Name:	<input type="text"/>
*Phone Number: (Please enter your 10-digit phone number i.e. xxx-xxx-xxxx)	<input type="text"/>
*Best time to call:	<input type="text"/>
*Time Zone:	<input type="button" value="Select"/>
*Email:	<input type="text"/>
Preferred Method of Enrollment:	<input type="button" value="Select"/>

If your preferred enrollment method is mail-based, please provide your mailing address below. (Street address, City, State, Zip code, Country)

Street Address:	<input type="text"/>
City/Town:	<input type="text"/>
State:	<input type="button" value="Choose Your State Here"/>
Postal Code:	<input type="text"/>
Country:	<input type="text"/>
*Referred by (Name of Organization/Healthcare Provider):	<input type="text"/>

Online Enrollment

Step 2:

- Participant logs in with their username & password
- Participant reviews Informed Consent and completes questionnaire

*CoRDS contacts participants annually to update their information. Participants may login anytime to update their information

The screenshot shows the Sanford Research Participant Portal login interface. At the top is the Sanford Research logo. Below it is a welcome message: "Welcome to the Sanford Research Participant Portal!". A list of three red square bullet points provides important information: "Username and Password are case sensitive.", "Pop-up blockers must be disabled for this application.", and "The Participant Portal will be unavailable each Sunday morning from 1:00am CST to 2:00am CST.". The main login area features a white box with "Username:" and "Password:" labels, each followed by a text input field. A red "Login" button is positioned below the password field. To the left of the login box, the text "Copyright © 2012 Velos Inc. All Rights Reserved." is visible. In the bottom right corner of the login box area, the version "v9.1.0 Build#650" is displayed. At the very bottom of the page, there is a link "Questions or Issues?" accompanied by an envelope icon.

SANFORD
RESEARCH

Welcome to the **Sanford Research** Participant Portal!

- Username and Password are case sensitive.
- Pop-up blockers must be disabled for this application.
- *The Participant Portal will be unavailable each Sunday morning from 1:00am CST to 2:00am CST.*

Username:

Password:

Login

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v9.1.0 Build#650

Questions or Issues?

Informed Consent

Sanford Health (US) | https://researchconnect.sanfordresearch.org:8443/velos/jsp/previewPortal.jsp?portalId=8

CoRDS Registry

Coordination of Rare Diseases at Sanford

Welcome to the CoRDS Registry Participant Portal!

➔ **Step 1: Please read the Patient Informed Consent below carefully**

The box below contains the CoRDS Assent and Consent forms.

- If you are a **Parent or Legally Authorized Representative**, please read the assent to whomever you are enrolling. And click [here](#) to print a copy for your records.
- For **Adults, Parents, and Legally Authorized Representatives**, please read the Consent Form below, and click [here](#) to print a copy for your records.

CONSENT

Affected Adults, Parents/Guardians, Legally Authorized Representatives Consent Form
Participation in Research for Affected Adults/Affected Minors
Sanford Research

Protocol Title: Coordination of Rare Diseases at Sanford (CoRDS)
Investigator: David Pearce, PhD
Department: Sanford Childrens Health Research Center

➔ **Step 2: Please complete the Patient Questionnaire(s):**

Please complete your questionnaire below. If you would like detailed instructions on how to complete your enrollment, click [here](#).

Your Forms:
[CoRDS Patient Questionnaire](#)

Tell us what you think!

We want to make your experience better! Please fill out the optional survey to let us know what you thought of the enrollment process.

[CoRDS Enrollment Survey](#)

What happens next?

- You will be contacted annually to update your information
- You may hear from us sooner if a researcher wants to recruit you into a study or clinical trial

Questions? Contact CoRDS:
877-658-9192
Cords@sanfordhealth.org

Cornelia de Lange Syndrome Questionnaire

to consent, we ask that you read to them, or have them read, the following information before you enroll them in the CoRDS registry. If your child/the participant does not agree to participate, you may not enroll them in the CoRDS Registry.



Step 2: Please complete the Patient Questionnaire(s):

The Cornelia de Lange Syndrome (CdLS) Registry is broken up into several forms, allowing you to answer some or all of the questions in one sitting.

If you would like detailed instructions on how to complete your enrollment, click [here](#). A glossary of medical terms can be found below.

For best results, use Internet Explorer or Mozilla Firefox to login to the secure web portal. **In the past, participants have encountered problems while completing the form with Google Chrome.** If you typically use Google Chrome as your web browser, we would recommend that you switch to Internet Explorer or Mozilla Firefox to access the portal.

Your Forms:

[CoRDS Enrollment Form](#)
[CdLS Registry Form 1 of 5](#)
[CdLS Registry Form 2 of 5](#)
[CdLS Registry Form 3 of 5](#)
[CdLS Registry Form 4 of 5](#)
[CdLS Registry Form 5 of 5](#)

[CdLS Questionnaire Glossary](#)

Tell us what you think!

We want to make your experience better! Please fill out the optional survey to let us know what you thought of the enrollment process.

[CoRDS Enrollment Survey](#)

CoRDS Enrollment Form

Preview

Form Name: CoRDS Enrollment Form

CoRDS Registry
Cooperation of Rare Diseases
& Sanford

CdLS Foundation
Cornelia de Lange Syndrome Foundation, Inc.

Today's Date *

INSTRUCTIONS

Thank you for taking the time to enroll with the CoRDS Registry for the CdLS Foundation. This questionnaire:

- Takes 10 - 20 minutes to complete
- Will refer to the person with the rare or unknown diagnosis as "the participant."
- Can be updated at any time by logging in to the CoRDS Participant Portal, or by contacting CoRDS personnel

Once per year, CoRDS will ask you to update your questionnaire.

If you have any questions while completing this form, please contact CoRDS at (877) 658 - 9192 during business hours, 8:30am-5:00pm (CST) Monday through Friday. If you need assistance after business hours, please leave a message or email cords@sanfordhealth.org

Who is completing this questionnaire? *

☐ Adult Participant: Participant is over the age of 18 and enrolling independently

☐ Parent/Guardian of Participant: Participant is under the age of 18

☐ Legally Authorized Representative (LAR) of Participant: Participant is over the age of 18 but is not cognitively or physically able to enroll

PERMISSIONS & DATA SHARING

By participating in CoRDS, your de-identified information will be visible to researchers who access the CoRDS Registry. Below are options that allow you to share your data with other entities; in the following questions, please select how you want your data shared. Please complete this section before moving on.

I give permission to CoRDS to contact me about participating in future research studies.*

☐ Yes ☐ No ☐ Don't Know

I give permission to CoRDS to contact me about donating a sample of blood, tissue, or other biospecimen for research in the future.*

☐ Yes ☐ No ☐ Don't Know

I give permission to CoRDS to provide a subset of de-identified information to other patient registries collecting information on rare diseases.*

☐ Yes ☐ No

I give permission to CoRDS to provide my information that may or may not be identifiable to the following Patient Advocacy Group(s) (PAGs) for non-research purposes.*

☐ Cornelia de Lange Syndrome Foundation ☐ None

PARTICIPANT INFORMATION

First Name: *

Middle Name:

Last Name: *

Primary Telephone Number: *

Email Address: *

CdLS Registry Form 1 of 5

Preview

Form Name: CdLS Registry Form 1 of 5

CoRDS Registry
Coordination of Rare Diseases at Sanford

CdLS Foundation
Cornelia de Lange Syndrome Foundation, Inc.

This is CdLS Registry Form 1 of 5

Today's Date * 01/15/2015

Diagnosis/Assessments

Has the participant ever had genetic testing?

☐ Yes

☐ No - Genetic testing was offered but the patient has not been tested because it cost too much

☐ No - Genetic testing was offered but the patient/guardian chose not to do it

☐ No - Genetic testing was offered but the patient has not done it yet for other reasons

☐ No - The healthcare provider has not talked about genetic testing for the patient

☐ No - The healthcare provider said the patient does not need genetic testing

☐ Unsure

☐ No - Other

If No - Other, please specify:

Which types of DNA (molecular) genetic tests have been performed? (Select all that apply.)

☐ CdLS gene panel

☐ Mitochondrial analysis

☐ Unsure

☐ Familial mutation analysis (Custom or targeted mutation test)

☐ Common mutation panel

☐ None

☐ Gene sequencing

☐ Exon/gene deletion/duplication panel

☐ Other

If other, please specify:

Which types of biochemical genetic tests have been performed? (Select all that apply.)

☐ Analyte, blood

☐ Analyte, urine

☐ Enzyme analysis

☐ Unsure

☐ None

☐ Other

If other, please specify:

Please select genetic change.

☐ NIPBL Mutation

☐ SMC1A Mutation

☐ SMC3 Mutation

☐ RAD21 Mutation

☐ HDAC8 Mutation

☐ NIPBL deletion

☐ SMC1A deletion

☐ SMC3 deletion

☐ RAD21 deletion

☐ HDAC8 deletion

☐ No change found

☐ Unsure

☐ Did not have testing

CdLS Registry Form 2 of 5

Preview

Form Name: CdLS Registry Form 2 of 5

CoRDS Registry
Coordination of Rare Diseases at Sanford

CdLS Foundation
Cornelia de Lange Syndrome Foundation, Inc.

This is CdLS Registry Form 2 of 5

Today's Date *

Heart/Cardiovascular

Was the participant born with a heart defect?

☐ Yes ☐ No ☐ Unsure

Has a doctor ever diagnosed the participant with any of the following? (Select all that apply)

☐ High blood pressure
☐ Low blood pressure

For the following congenital heart defects, select the status and the age of diagnosis. (Select all that apply.)

Aortic Valvar Stenosis (AS)

☐ Currently a problem ☐ Not a problem today, but was in the past ☐ Never a problem ☐ Unsure

Age at Diagnosis ☐ Unsure Years: Months:

Aortic valve regurgitation

☐ Currently a problem ☐ Not a problem today, but was in the past ☐ Never a problem ☐ Unsure

Age at Diagnosis ☐ Unsure Years: Months:

Atrial Septal Defect (ASD)

☐ Currently a problem ☐ Not a problem today, but was in the past ☐ Never a problem ☐ Unsure

Age at Diagnosis ☐ Unsure Years: Months:

Atrioventricular Septal Defect (or AV Canal Defect)

☐ Currently a problem ☐ Not a problem today, but was in the past ☐ Never a problem ☐ Unsure

Age at Diagnosis ☐ Unsure Years: Months:


Coarctation of the Aorta

☐ Currently a problem ☐ Not a problem today, but was in the past ☐ Never a problem ☐ Unsure

CdLS Registry Form 3 of 5

Preview

Form Name: CdLS Registry Form 3 of 5

This is CdLS Registry Form 3 of 5

Today's Date *

Neurology

In the past, has the participant experienced any of the following neurological symptoms? (Select all that apply.)

<input type="checkbox"/> Anxiety	<input type="checkbox"/> Arm or leg weakness	<input type="checkbox"/> Coordination problems
<input type="checkbox"/> Dizziness or vertigo	<input type="checkbox"/> Headache	<input type="checkbox"/> Hearing problems
<input type="checkbox"/> Hiccups	<input type="checkbox"/> Numbness	<input type="checkbox"/> Pain in spine or limbs
<input type="checkbox"/> Seizure	<input type="checkbox"/> Vision problems	<input type="checkbox"/> None
<input type="checkbox"/> Unsure	<input type="checkbox"/> Other	

If other, please specify:

For the following neuromuscular or movement disorders, select the status and the age of diagnosis. (Select all that apply.)

Ataxia

☐ Currently a problem ☐ Not a problem today, but was in the past ☐ Never a problem ☐ Unsure

Age at Diagnosis: ☐ Unsure Years: Months:

Ballismus

☐ Currently a problem ☐ Not a problem today, but was in the past ☐ Never a problem ☐ Unsure

Age at Diagnosis: ☐ Unsure Years: Months:

Bradykinesia

☐ Currently a problem ☐ Not a problem today, but was in the past ☐ Never a problem ☐ Unsure

Age at Diagnosis: ☐ Unsure Years: Months:

Chorea



☐ Currently a problem ☐ Not a problem today, but was in the past ☐ Never a problem ☐ Unsure

Age at Diagnosis: ☐ Unsure Years: Months:

CdLS Registry Form 4 of 5

Preview

Form Name: CdLS Registry Form 4 of 5

This is CdLS Registry Form 4 of 5

Today's Date *

Eyes/Vision

Has the participant had any eye surgeries or implants?

☐ Yes ☐ No ☐ Unsure

Has the participant been diagnosed with any of the following eye or vision conditions? (Select all that apply.)

<input type="checkbox"/> Amblyopia	<input type="checkbox"/> Anatomical Blindness	<input type="checkbox"/> Astigmatism
<input type="checkbox"/> Cataract	<input type="checkbox"/> Color blindness	<input type="checkbox"/> Conjunctival Abnormality
<input type="checkbox"/> Cortical Blindness (Visual Field Loss)	<input type="checkbox"/> Curly eyelashes	<input type="checkbox"/> Depth perception problems
<input type="checkbox"/> Farsighted	<input type="checkbox"/> Glaucoma	<input type="checkbox"/> Lacrimal (tear duct) blockage
<input type="checkbox"/> Long eyelashes	<input type="checkbox"/> Nearsighted	<input type="checkbox"/> Nystagmus (involuntary rapid eye movement)
<input type="checkbox"/> Ptosis (droopy eyelid)	<input type="checkbox"/> Retinal detachment from near sightedness	<input type="checkbox"/> Retinal detachment from trauma
<input type="checkbox"/> Strabismus (eye turned in or out)	<input type="checkbox"/> Synophrys (joined eyebrows)	<input type="checkbox"/> None
<input type="checkbox"/> Unsure	<input type="checkbox"/> Other	

If other, please specify:

Which of the following eye or vision devices does the participant use to help with vision? (Select all that apply.)

<input type="checkbox"/> Contacts	<input type="checkbox"/> Glasses	<input type="checkbox"/> Magnifier
<input type="checkbox"/> None used	<input type="checkbox"/> Unsure	<input type="checkbox"/> Other

If other, please specify:

Puberty

For female participants, what is the age of onset of menses (age at first period)?

☐ Unsure ☐ Has not menstruated

Years: Months:

At what age did the following pubertal changes start?

Underarm hair	Years: <input type="text" value="Select an Option"/>	Months: <input type="text" value="Select an Option"/>	<input type="checkbox"/> Unsure
Facial hair	Years: <input type="text" value="Select an Option"/>	Months: <input type="text" value="Select an Option"/>	<input type="checkbox"/> Unsure
Breast development	Years: <input type="text" value="Select an Option"/>	Months: <input type="text" value="Select an Option"/>	<input type="checkbox"/> Unsure

CdLS Registry Form 5 of 5

Preview

Form Name: CdLS Registry Form 4 of 5

CoRDS Registry
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This is CdLS Registry Form 4 of 5

Today's Date * 01/15/2015

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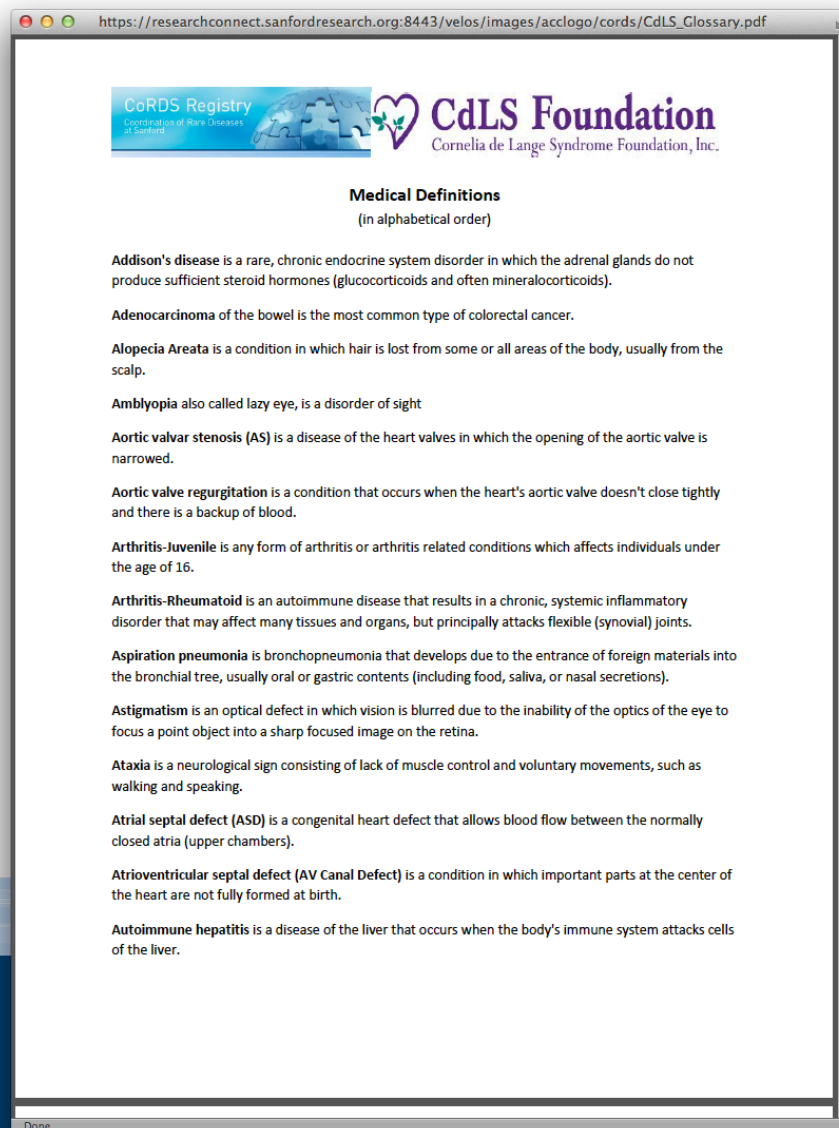
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At what age did the following pubertal changes start?

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Facial hair	Years: <input type="text"/>	Months: <input type="text"/>	<input type="checkbox"/> Unsure
Breast development	Years: <input type="text"/>	Months: <input type="text"/>	<input type="checkbox"/> Unsure

CdLS Questionnaire Glossary



What information is collected in the CoRDS module?

Demographic information:

- Name
- Date of birth
- Gender
- Race
- Contact information
- Data-sharing preferences



*Includes Common Data Elements (CDEs) as recommended by the National Institute of Health (NIH).

What information is collected in the disease-specific modules?

Clinical diagnosis information

- Diagnosis
- Symptoms
- When/where testing occurred
- How diagnosis was made
- Family history



Data Management



- Velos E-Research Software
- Centralized, relational database of rare disease data
- Provides a mechanism to securely collect, modify, display, & report data
- Secure patient portal allows for online enrollment
- Easily searchable using common data query (SQL) commands
- Supports multiple languages, data curation



CoRDS Partners



Lowe Syndrome Association

FMD Chat



BATTEN DISEASE
SUPPORT & RESEARCH
ASSOCIATION
A LIGHT IN A WORLD OF DARK



madisons
FOUNDATION



XLH
NETWORK



SANFORD
Children's



ring14 USA
OUTREACH



SANFORD
RESEARCH

CoRDS Registry

Coordination of Rare Diseases
at Sanford



Questions?

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Catherine.Olson@sanfordhealth.org

www.sanfordresearch.org/cords

