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:	Benjamin Forred
Department:	Sanford Children's Health Research Center

Instructions: If you have previously completed a CoRDS Informed Consent Form, please read carefully as the terms may have changed.

The current Informed Consent Form and Assent Form are available on the CoRDS website (http://www.sanfordresearch.org/cords/), online within the online portal, and via mail, email, or telephone per participant request. CoRDS encourages participants to review the Informed Consent Form and Assent Form regularly for any revisions. It is your responsibility to ensure you have read the current Informed Consent Form and Assent Form.

What is the purpose of this study?

The purpose of the Coordination of Rare Diseases at Sanford (CoRDS) is to establish an international rare disease patient registry. CoRDS' goal is to collect and provide a valuable resource of information, and to accelerate patient recruitment into research studies and clinical trials.

Who can participate?

Any individual with a rare, uncommon, or undiagnosed disease, as well as those who are unaffected carriers are welcome to enroll in the CoRDS Registry. A parent or guardian may enroll a child who is under the age of 18. A Legally Authorized Representative (LAR) may enroll an adult over the age of 18 who is unable to consent.

What will happen in this study?

If you choose to enroll, you will be asked to complete a brief questionnaire(s). The questionnaire(s) requests your basic contact, socio-demographic, and health information, as well as your communication and research preferences. This information will be saved under a unique coded identifier. CoRDS will send a reminder if your questionnaire has not been completed, and will send an annual reminder to update your information or confirm that it is up to date.

Sometimes, it is important for researchers to know how many members of a family are affected by a given condition and how diseases are inherited within a family. You will have an option to link your account with other family members who are enrolled in CoRDS. By linking your account to a family member's account, you are agreeing to have your name, date of birth, city/state residence, and family relation shown to the family member to which you would like to be linked. You will not be able to look at or edit each other accounts. Researchers will not be able to identify who the individuals are; rather they will only know that you are related to the family member you have linked to.

Is this study voluntary?

Participation in CoRDS is voluntary. You are free to withdraw at any time, for any reason. To withdraw, please contact CoRDS by mail or email and select one of the following options.

- 1. Leave identifiable information in CoRDS, but do not contact me again;
- 2. Remove identifiable information and leave de-identified information in CoRDS;
- 3. Remove all information from CoRDS.

You will have 30 days to decide what you wish to do with your data. CoRDS will contact you to confirm that your data has been modified/deleted according to your wishes.

If CoRDS does not hear from you within 30 days, your identifiable information will be removed and data will be stored in the registry indefinitely.

What are the benefits of this study?

There are no direct benefits to participating in CoRDS but the data you contribute may help advance research for rare diseases which would benefit a wider population. By participating in CoRDS you may be contacted about research opportunities you qualify for, and can decide at that time if you would like to participate, however CoRDS cannot guarantee that a researcher will request to contact you.

Is the information kept confidential? What are the risks of participation?

CoRDS will make every effort to keep all information gathered in the registry confidential, but this cannot be guaranteed. There is a minimal risk associated with the loss of confidentiality.

How will my information be accessed? Can I choose how my information is shared?

On the questionnaire(s), you can choose how you would like CoRDS to share your information. Information in CoRDS may be accessed in the following ways:

- Researchers may access de-identified information once they are approved by the CoRDS Scientific Review Committee. If you enter a disease-specific registry for a Patient Advocacy Group (PAG), that PAG may ask to be involved in the review process. If a researcher wishes to notify you about a research opportunity, CoRDS Personnel will contact you on their behalf. There is no obligation to participate – it is always up to you to contact the researcher. CoRDS ensures that all research study referrals have been reviewed and approved by an IRB as applicable.
- 2. A subset of de-identified information may be shared with certain other databases in order to avoid a duplication of efforts and to increase knowledge and understanding of rare diseases.
- Patient Advocacy Groups (PAGs) representing individuals with rare or uncommon diseases may have access to information that may or may not be identifiable for non-research purposes. These PAGs have signed a contract stating they will not use the information for research purposes.

Who should I contact if I have questions?

- For general questions about CoRDS or enrollment: call (877) 658-9192 or email cords@sanfordhealth.org
- If you feel you have been harmed: Benjamin Forred, (605) 312-6416 If you have questions about your rights as a research participant: Sanford Health Institutional Review Board (IRB), (605) 312-6430



Instructions

Thank you for taking the time to enroll with the CoRDS/Hypersomnia Registry. This module will ask you questions specific to your diagnosis. The questions below were developed in partnership with the Hypersomnia Foundation. Please note, this module:

- Takes approximately 1 hour to complete
- Will refer to the person with the diagnosis as "the participant"
- References the participant's genetic report
- Can be updated at any time by logging into the CoRDS online portal or by contacting CoRDS personnel

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

Permissions & Data Sharing

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

□ Hypersomnia Foundation

□ I do not give my permission

SLEEP DURATION & FREQUENCY

1. When (date OR at what age) did the participant first start feeling as if they were excessively sleepy or needed more sleep than most people?

Date:

Age: _____

2. When (date OR at what age) did the participant (or their parent(s)/guardian(s)/LAR) first seek help for their excessive sleepiness or need for sleep?

Date:

Age:_____

3. What is the longest the participant has ever slept at one time while TAKING MEDICATION for their excessive sleepiness or need for sleep (please round to the nearest hour)?

4. What is the longest the participant has ever slept at one time while NOT TAKING MEDICATION for their excessive sleepiness or need for sleep (please round to the nearest hour)?

	Hours						
5.	5. How often does the participant have difficulty waking up for the day? Please select ONE response.						
	□ Never	1-2 times a week					
	□ Less than once a month	\Box 3-4 times a week					
	Less than once a week	Daily or almost daily					
6.	6. How often does the participant usually feel rested after waking up for the day? Please select O response.						
	Daily or almost daily	□ 1-3 times a month					
	□ 3-4 times a week	Less than once a month					
	1-2 times a week	□ Never					
7.	How long does a typical nap last for the partic	ipant (hours and/or minutes)?					
	Hours:	Minutes:					
8.	After a nap, how does the participant typically	r feel? Please select ONE response.					
	Not refreshed	□ Refreshed					
	Somewhat refreshed						

SLEEP SYMPTOMS

When answering questions 9 and 10, please think back to when the participant's symptoms WERE THE WORST they have ever been.								
9. Was the participant taking medication to treat their excessive sleepiness or need for sleep when the symptoms were the worst they have ever been?								
□ Yes] No				
10. When the SYMPTOMS WERE THE WORST they have ever been, how often did the participant experience each of these symptoms? Please select ONLY ONE response for each row.								
Symptom	Never	Once or twice in my life	Once or twice a year	Monthly	Once or twice a month	Weekly	Every day	More than once a day

[1	1	 1	
Excessive daytime sleepiness					
Long sleep (sleeping longer than 10 hours at a time)					
Intentional nap (sleeping more than once in a 24-hour period)					
Fall asleep during the day without meaning to					
Use more than one alarm device to wake up					
Trouble waking up (sleep inertia) and functioning with normal alertness (sleep drunkenness)					
'Brain fog' (unable to think clearly or concentrate at any time throughout the day)					
Difficulty remembering things					
Weakness or slackness in the jaw, face, or neck with laughter or strong emotions such as anger, fear, stress, or excitement					
Sleep paralysis (being unable to move when falling asleep or waking up)					
Night sweats					
Restless sleep					
Hypersexuality					
Hyperphagia (Abnormally increased appetite and ingestion of food [typically junk food])					
Hypnagogic hallucinations (seeing or hearing things that aren't really there) when falling asleep					

Hypnopompic hallucinations								
(seeing or hearing things that aren't really there) when waking up								
Automatic behavior (doing something without realizing/being aware) - Automatic behavior usually happens when sleepy but trying to stay awake (e.g., arriving at a destination without being able to recall how)								
When answering questions 11 a	nd 12, plea	ase think	back ove	r the pas	st 30 days	5		
11. Has the participant been sleepiness or need for sleepiness or ne								'e
□ Yes		No			No medication prescribed			
12. Over the PAST 30 DAYS, symptoms? Please select				-	enced ea	ch of the t	following	
						¥		۲.
Symptom			Never	Once or twice	3 or 4 times	2-6 times each week	Every day	More than once a day
Symptom Excessive daytime sleepiness			Never		or 4 tiı	S	Every day	More than once a da
	10 hours a	ıt a	Never		or 4 tiı	S	Every day	More than once a da
Excessive daytime sleepiness Long sleep (sleeping longer than			Never		or 4 tiı	S	Every day	More than once a da
Excessive daytime sleepiness Long sleep (sleeping longer than time) Intentional nap (sleeping more tl	han once ir	n a 24-	Never		or 4 tiı	S	Every day	More than once a da

	1			
Trouble waking up (sleep inertia) and functioning with normal alertness (sleep drunkenness)				
'Brain fog' (unable to think clearly or concentrate at any time throughout the day)				
Difficulty remembering things				
Weakness or slackness in the jaw, face, or neck with laughter or strong emotions such as anger, fear, stress, or excitement				
Sleep paralysis (being unable to move when falling asleep or waking up)				
Night sweats				
Restless sleep				
Hypersexuality				
Hyperphagia (Abnormally increased appetite and ingestion of food [typically junk food])				
Hypnagogic hallucinations (seeing or hearing things that aren't really there) when falling asleep				
Hypnopompic hallucinations (seeing or hearing things that aren't really there) when waking up				
Automatic behavior (doing something without realizing/being aware) - Automatic behavior usually happens when sleepy but trying to stay awake (e.g., arriving at a destination without being able to recall how)				

SLEEP SCHEDULE							
Please answer the following questions as they relate to a workday (which would include paid employment, volunteer work, school, or other obligations that require the participant to be out of bed at a specific time) or non-workday (any day in which the participant does not have to be up at a specific time).							
13. What time does the participant usually go to bed on workdays (ONLY SELECT either AM/PM)?							
i	AM	OR	::	PM			
14. What time does the second se	ne participant us	sually wake up on	workdays (ONLY SE	LECT ONE AM/PN	1)?		

·	AM	OR	;	PM
15. What time does t AM/PM)?	the participant us	sually get out of bec	l for their workday	(ONLY SELECT ONE
;;	AM			
OR			•	PM
16. How long does th	ne participant usu	ally sleep (NOT jus	t lying in bed) at ni	ght on workdays?
Hours:			Minutes:	
17. What time does t	the participant us	sually go to bed on	non-workdays? (O	NLY SELECT ONE AM/F
	AM		_	
OR			:	PIVI
18. When does the p	articipant usually	/ wake up on non-w	vorkdays? (ONLY S	ELECT ONE AM/PM)
::::	AM		::	PM
-	the participant us	sually get out of bec	l for their non-wor	kday? (ONLY SELECT C
	AM		::	PM
()R				
OR 20. How long does th	ne participant usu			
20. How long does th	ne participant usu Hours			
20. How long does th	Hours	ually sleep (NOT jus	t lying in bed) at ni 	ght on non-workdays?
20. How long does the average of a second se	Hours	ually sleep (NOT just	t lying in bed) at ni 	ght on non-workdays?

PHYSICIAN/MEDICAL PROFESSIONALS

22. How many doctors or medical professionals has the participant seen while trying to get a diagnosis/treatment for their excessive sleepiness or need for sleep? (0-25)

23. What type of doctor or medical professional diagnosed the participant's hypersomnia (gave a name to their excessive sleepiness or need for sleep)? Please select all that apply.					
 Not applicable – participant's excessive sleepiness or need for sleep has never been diagnosed. If not applicable, please skip to question 24. 					
Nurse practitioner	General psychiatrist				
Physician assistant	Sleep doctor - Neurologist				
General internist	Sleep doctor - Psychologist				
Family physician	Sleep doctor – Psychiatrist				
General neurologist	Sleep doctor – Pulmonary/lung specialist				
	Sleep doctor – Specialty Unknown				
Other medical professional (please specify below)					
If "Other medical professional", please specify:					

DIAGNOSES

24. Please indicate whether any of the following disorders were a diagnosis the participant had in the past or the participant currently has and whether the participant's doctor indicated that this disease or disorder was the cause of the participant's excessive sleepiness or need for sleep.

Disease or disorder	Never been diagnosed	Diagnosed in the past, but no longer a diagnosis	Current diagnosis	Doctor said this was/is a cause of the participant's excessive sleepiness or need for sleep
Acromegaly				
Alzheimer disease				
Diabetes (type 1 or 2)				
Dysautonomia				
Hypothyroidism				

Myotonic dystrophy				
Obesity				
Parkinson disease				
Postural orthostatic				
tachycardia				
25. Please indicate whethe past or the participant or disorder was the cau	currently has and	whether the particip	pant's doctor inc	licated that this disease
Disease or disorder	Never been diagnosed	Diagnosed in the past, but no longer a diagnosis	Current diagnosis	Doctor said this was/is a cause of the participant's excessive sleepiness or need for sleep
Alcoholism				
Anxiety				
Attention deficit disorder (ADD)/Attention-deficit hyperactivity disorder (ADHD)				
Bipolar disorder				
Depression				
Seasonal affective disorder				
Substance abuse (including prescribed medication or other substances)				
26. Please indicate whethe past or the participant or disorder was the cau	currently has and	whether the particip	pant's doctor inc	licated that this disease
Disease or disorder	Never been diagnosed	Diagnosed in the past, but no longer a diagnosis	Current diagnosis	Doctor said this was/is a cause of the paticipant's excessive sleepines or need for sleep
Connective tissue disorder				

Ehlers-Danlos syndrome		
Fibromyalgia		
Systemic exertion intolerance disease (also known as chronic fatigue syndrome, myalgic encephalopathy or myalgic encephalomyelitis)		

27. Please indicate whether any of these circadian rhythm disorders were a diagnosis the participant had in the past or the participant currently has and whether the participant's doctor indicated that this disease or disorder was the cause of the participant's excessive sleepiness or need for sleep.

Disease or disorder	Never been diagnosed	Diagnosed in the past, but no long a diagnosis	Current	Doctor said this was/is a cause of the participant's excessive sleepiness or need for sleep
Advanced sleep phase syndrome				
Delayed sleep phase syndrome				
Non-24-hour sleep-wake disorder				
Shift work sleep disorder				
28. Please indicate whetl past or the participan or disorder was the c	nt currently has and	whether the particip	ant's doctor indi	cated that this disease
Disease or disorder	Never been diagnosed	Diagnosed in the past, but no longer a diagnosis	Current diagnosis	Doctor said this was/is a cause of the participant's excessive sleepiness or need for sleep
Brain tumor				
Encephalopathy				

Disease or disorder	Never been diagnosed	Diagnosed in the past, but no longer a diagnosis	Current diagnosis	Doctor said this was/is a cause of the participant's excessive sleepiness or need for sleep
30. Please indicate whethe the participant currentl disorder was the cause	y has and whethe	er the participant's d	loctor indicated	that this disease or
Narcolepsy; Type not specified				
Narcolepsy; Type 2 (without cataplexy or hypocretin deficiency)				
Narcolepsy; Type 1 (with cataplexy or hypocretin deficiency)				
Kleine-Levin syndrome				
Idiopathic hypersomnia				
Hypersomnia (not otherwise specified)				
Disease or disorder	Never been diagnosed	Diagnosed in the past, but no longer a diagnosis	Current diagnosis	Doctor said this was/is a cause of the participant's excessive sleepiness or need for sleep
29. Please indicate whethe had in the past or the p this disease or disorder	articipant current	ly has and whether	the participant's	s doctor indicated that ss or need for sleep.
Hydrocephalus				
Head trauma (concussion, whiplash, traumatic brain injury, loss of consciousness)				
Headaches; Migraine				
Headaches; Chronic				
Epilepsy or seizures				

Acting out dreams during sleep (REM sleep behavior disorder)				
Body rocking				
Head banging during sleep (jactatio capitis nocturna)				
Sleep talking (somniloquy)				
Sleep walking (somnambulism)				
31. Please indicate whether past or the participant c or disorder was the caus	urrently has and wh	ether the participa	ant's doctor indica	ted that this disease
Disease or disorder	Never been diagnosed	Diagnosed in the past, but no longer a diagnosis	Current diagnosis	Doctor said this was/is a cause of the participant's excessive sleepiness or need for sleep
Insufficient sleep (that is, the participant just needs to sleep more)				
Periodic limb movements of sleep				
Restless legs syndrome				
32. Please indicate whether the past or the participa disease or disorder was	int currently has and	l whether the part	icipant's doctor in	dicated that this
Disease or disorder	Never been diagnosed	Diagnosed in the past, but no longer a diagnosis	Current diagnosis	Doctor said this was/is a cause of the participant's excessive sleepiness or need for sleep
Sleep apnea; Central				
Sleep apnea; Obstructive				

Sleep apnea; Mixed		
Upper airway resistance syndrome		

33. If the participant's sleepiness or excessive need for sleep began after an infection, please select the most appropriate responses below.

Infection	Never been diagnosed	Diagnosed in the past, but no longer a diagnosis	Current diagnosis	Doctor said this was/is a cause of the participant's excessive sleepiness or need for sleep
Epstein-Barr virus				
Encephalitis				
Encephalitis lethargica (sleepy sickness)				
Guillain-Barre syndrome				
H1N1/Swine flu				
HIV/AIDS				
Meningitis				
Mononucleosis or mono				
Whipple's disease				
Other infection (specify below)				

If "Other infection", please specify:

34. Please indicate whether any of these types of strokes were a diagnosis the participant had in the past or the participant currently has and whether the participant's doctor indicated that this disease or disorder was the cause of the participant's excessive sleepiness or need for sleep.

Disease or disorder	Never been diagnosed	Diagnosed in the past, but no longer a diagnosis	Current diagnosis	Doctor said this was/is a cause of the participant's excessive sleepiness or need for sleep
Stroke; Bleed (hemorrhagic)				

Stroke; Clot (thrombotic or embolic)					
Stroke; Don't know					
35. If the participant's sleepiness or excessive need for sleep began after a diagnosis that is not listed, please specify below:					
36. Did the participant's excessive sleepiness or ne vaccination?	eed for sleep begin shortly after receiving a				
□ Yes	□ No				
If "Yes", please select all that apply:					
Human papillomavirus (HPV)	Seasonal influenza (flu)				
Meningococcal meningitis	Other vaccine (specify below)				
If "Other vaccine", please specify:					

MEDICATION

37. What stimulant medications has the participant been prescribed by a medical doctor or other medical professional for their excessive sleepiness or need for sleep? Please select ONY ONE response for each row.

Medication	Never Taken	Taken in the past, but not taking now	Currently taking at least once a week but less than every day	Currently taking every day or almost every day
Dexmethylphenidate (Focalin)				
Dextroamphetamine (Dexedrine)				
Lis-dexamfetamine (Vyvanse)				
Mazindol				

Methylphenidate (Ritalin, Metadate, Concerta, Daytrana)					
Mixed amphetamine salts (Adderall)					
Pemoline (Betanamin, Cylert, Tradon, and Ceractiv)					
If the participant's stimula currently taking this medic		· • •	ify name(s) and w	/hether	the participant is
Medication	Never Taken	n in the past, not taking now	Currently taking least once a we but less than ev day	ek	Currently taking every day or almost every day
38. What wakefulness other medical pro response for each	fessional for the				edical doctor or e select ONLY ONE
Wakefulness-promoting agent	Never Taken	n in the past, not taking now	Currently taking least once a we but less than ev day	ek	Currently taking every day or almost every day
Armodafinil (Nuvigi)					
Modafinil (Provigil)					
If the participant's wakefu participant is currently tak		 · •		(s) and	whether the
Other wakefulness- promoting agent	Taken in the pa but not taking	Currently takin a week but les day	ng at least once s than every		ntly taking every r almost every day

39. What sleeping pills (sedative/hypnotics) has the participant been prescribed by a medical doctor or other medical professional for their excessive sleepiness or need to sleep? Please select ONLY ONE response for each row.

response for eac	h row.					
Sleeping pill (sedative/hypnotics)	Never Taken	pa	ken in the st, but not king now	Currently taking at least once a week b less than every day		Currently taking every day or almost every day
Alprazolam (Xanax)						
Clonazepam (Klonopin)						
Diazepam (Valium)						
Doxepin (Silenor)						
Eszopiclone (Lunesta)						
Ramelteon (Rozerem)						
Suvorexant (Belsomra)						
Temazepam (Restoril)						
Trazodone (Desyrel, Oleptro)						
Triazolam (Halcion)						
Zaleplon (Sonata)						
Zolpidem (Ambien, Ambien CR, Intermezzon, Stilnox, Stilnoct, Sublinox, Hypnogen, Zonadin, Sanval, Zolsana)						
If the participant's sedat participant is currently ta					s) an	d whether the
Other Sleeping pill (sedative/hypnotics)	Taken in the past, but not taking nov		-	king at least once a ss than every day		rrently taking every / or almost every /

40. What antidepressant m prescribed for the parti ONE response for each	cipant for th		or or other medical profes piness or need to sleep?	
Antidepressant medication	Never Taken	Taken in the past, but not taking now	Currently taking at least once a week but less than every day	Currently taking every day or almost every day
Amitriptyline (Elavil)				
Bupropion (Wellbutrin)				
Citalopram (Celexa)				
Clomipramine (Anafranil)				
Duloxetine (Cymbalta)				
Escitalopram (Lexapro)				
Fluoxetine (Prozac)				
Fluvoxamine (Luvox)				
Phenelzine (Nardil)				
Protriptyline (Vivactil)				
Sertraline (Zoloft)				
Tranylcypromine (Parnate)				
Venlafaxine (Effexor)				

If the participant's antidepressant medication is not listed, please specify name(s) and whether the
participant is currently taking this medication and, if so, how often:

Other Antidepressant medication	Taken in the past, but not taking now	Currently taking at least once a week but less than every day	Currently taking every day or almost every day

41. What thyroid supplements has a medical doctor or other medical professional ever prescribed for the participant for their excessive sleepiness or need to sleep? Please select ONLY ONE response for each row.

Thyroid supplement	Never Taken	Taken in the past, but not taking now	Currently taking at least once a week but less than every day	Currently taking every day or almost every day
Armour Thyroid				
Levothyroxine (Synthroid)				
Liothyronine sodium (Cytomel)				

If the participant's thyroid supplement is not listed, please specify the name(s) and whether the participant is currently taking this medication and, if so, how often:

Other Thyroid supplement	Taken in the past, but not taking now	Currently taking at least once a week but less than every day	Currently taking every day or almost every day

42. What other medications has a medical doctor or other medical professional ever prescribed for the participant for their excessive sleepiness or need to sleep?

Other medication	Never Taken	Taken in th but not tak	-	Currently taking a least once a week but less than ever day		Currently taking every day or almost every day	
Atomoxetine (Strattera)							
Baclofen							
Clarithromycin (Biaxin)							
Clonidine (Catapres, Dapvay)							
Flumazenil							
Guanfacine (Intuniv <i>,</i> Tenex)							
Melatonin							
Sodium oxybate (Xyrem)							
If the participant's medication for their excessive sleepiness or need to sleep is not listed in any of these sections, please specify name(s) and whether the participant is currently taking this medication and, if so, how often:							
Other medication	Taken in the past, but not taking now		once a week but less than		ev	Currently taking every day or almost every day	
43. Please list all of the participant's current prescription medications (which WERE NOT USED TO TREAT THEIR EXCESSIVE DAYTIME SLEEPINESS OR NEED FOR SLEEP) and what condition the medication is used to treat.							
Other medication 1:		Condition for medication 1:					
Other medication 2:		Condition for medication 2:					

Other medication 3:	Condition for medication 3:		
44. What over-the counter and other non-pharmaceutical substances does the participant use to help treat their excessive sleepiness or need for sleep? Please select all that apply.			
Ephedrine			
Energy drinks			
🗌 Khat	Herbal or botanical products		
	Methamphetamine		
Melatonin	Other (please specify)		
If "Other", please specify:			

Thank you for your participation!

Questions? Contact CoRDS Personnel

Sanford Research | 2301 East 60th Street North | Sioux Falls, South Dakota 57104 **Phone** (toll-free): 1 (877) 658-9192 | **Email**: CoRDS@sanfordhealth.org