


Omics & Biospecimen WG Minimal for Core Protocol

Will be tailored a bit by age. Also will minimize number of tubes (maybe 3). At least once (e.g. not longitudinal) and in a hybrid fashion (e.g., tube, swab, tasso, remote vs in person, etc). Minimal collection = participant. Note, some sites are already successfully using the Tasso with 18m and 24m olds with DS. Cost was for all of this was discussed, some costs will fall on Biospecimen Core (materials, analysis), but others will be on sites (e.g., preprocessing). Still TBD and this will shape the final protocol for what will be on the minimal protocol.

Minimal



Data Collection Measure	Site	Collection method	Age Group	In-Person/Remote/Hybrid	Estimated Time to Complete	Cadence (One time, annually, etc.)
DNA (germline DNA WGS)		Tasso, PAXGene DNA	2+	Hybrid		Once
DNA (Microbiome)		Swab	0+	Hybrid		Once
RNA (whole blood)		Tasso (only if PAXGene RNA is not available), PAXGene RNA	2+	Hybrid		Once
Plasma		Purple top EDTA	2+	In-person		Once
Buffy coat		Purple top EDTA	2+	In-person		Once
RBC		Purple top EDTA	2+	In-person		Once
Fixed cells (CyTOF)		Purple top EDTA	2+	In-person		Once


- NOTE 1: still questions regarding infants and under 2 for blood draw volumes. We may need to make a Minimal collection that's tailored based on weight (for blood draw volumes).
- NOTE 2: There will still be room to collect additional samples beyond the common proposed here, as long as it doesn't prevent collection of the minimal/optimal data sets.
- NOTE 3: General frame work for costs is that the DS-Biorepository will cover costs for minimal and optimal sample collection materials, but wishlist and specimen acquisition costs are under the purview of the collection site. This is a great opportunity to apply for more funding.

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Medical Data & EHR WG Minimal for Core Protocol – In Person

Will take into account some visits may be remote for some studies. Ideally collect these measures at each visit. Will provide some training/SOP for consistency (e.g., head circ, body size) and could supplement from med records if within certain time window. Many measures won't be relevant for <2 yo. See 2nd slide p2

Minimal IN-PERSON




Data Collection Measure	Age Group	Est. Completion Time	Cadence (once, annually)	EHR complement	EHR substitute (time frame)	Additional Considerations
Length (2X)	0-3 y (record if L or Ht)	15 min (comfort & undressing)	Every visit	X	Age dependent	Decode-IT (n=500) Other remote only Length for non-standing
Height (2X)	Age> 3y		Every visit	X		
Weight (1X)	All		Every visit	X		
SBP	All	10-15 min	Every visit	X	≥12 y previous 12 mo <12y previous 6 mo	Automated; but do we do by auscultation if elevated
2 in one sitting; 3 if discordant	All		Every visit	X		
DBP	All		Every visit	X		
HR	All		Every visit	X		
SpO2	All	5 min	Every visit	X		Age<1mo Consider CHD, UE & LE
Head circumference	All	5-10 min	Age>18y once	X		
Waist (umbilicus)	Age>3y	5 min	Every visit			

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Regarding audiology, recognize that perhaps will need to be done at separate visit or taken from med records. Also in conversation about DXA scan, comment was made about trying to keep minimal common protocol 'at minimal risk' for purposes of IRB (e.g., so sedated MRIs would not be). Note, brain MRI was listed under 'wish' for >6 or perhaps to be gathered from EHR. BUT excellent discussion on this topic may have raised it to optimal.

Minimal IN-PERSON Cont.



Data Collection Measure	Age Group	Est. Completion Time	Cadence (once, annually)	EHR complement	EHR substitute (time frame)	Additional Considerations
Audiology	Subset Subset repeated	Age<5y 60 min; maybe longer	Ideally 2 X; alternating visits	X	Ok within 12 mo For individuals with rehab program—may not be in EHR	Neurodevelopment assessment Could be done on day of clinical appointment
Hand grip (3X each hand; effort dependent— take best of 3)	Age>6y	5-10 min	annually			
6-minute walk test	Age>6y subset	15-min	At least once, Annually for subset			
Pubertal stage • exam • self-assessment	Age>8y-18y	5-10 min Trained personnel	Annually until TS5 Up to age 18y	? Endocrine records		Remote if self- assessment w/direct observation by caregiver
Fracture Questionnaire	All	2-min	Annually	X Radiology Ortho Primary care		medical questionnaire, and if yes, complete fracture questionnaire


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Demographics, SES, etc WG

Minimal for Core Protocol

Keeping basic intake forms short (~10-15 min). Can be collected remotely. Plan to use NDAR GUIDs and will harmonize with DS Connect forms/Q as much as possible. Will try and keep PHI site/local versus in the DCC.

Minimal




Data Collection Measure	Age Group	In-Person/ Remote/ Hybrid	Estimated Time to Complete	Cadence (One time, annually, etc.)
Data Intake Form	All	All	10-15 minutes	Verify some questions annually
SDOH – Basic	All	All	3 minutes	Annually
GUID Form	All	All	2 minutes	One time
Study ID	All	All	2 minutes	One time
Total Time			<18 minutes	

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Assessment WG Minimal Core Protocol

Really focused on developmental/lifespan perspective. Took into account every project’s proposed batteries and looked for common domains to include for minimal. Not shown, but something for sensory got added to this list. Taking care to think about feasibility with DS participants meaning, successful admin and score distribution and appropriate floor/ceiling). Caregiver AND in person data collection planned. TBD where some data (e.g., sleep Q) should be accounted (here or under medical). Need to compile overall burden of this battery plus other things to be gathered per other WGs.


Minimal



Measure	Type	Age Group			In-Person/ Remote/ Hybrid	Estimated Time to Complete	Cadence (One time, annually, etc.)
		0-5	6-21	22+			
Vineland Adaptive Behavior Scales -3	Caregiver Rating	x	x	x	Hybrid	15 minutes	
Developmental Profile Scales 4	Caregiver Rating	x	x		Hybrid	30 minutes	
Survey of Well Being for Young Children	Caregiver Rating	x			Remote	10 minutes	
Rothbart IBQ/ECBQ/CBQ	Caregiver Rating	x	X		Hybrid	15 minutes	
Modified Checklist for Autism in Toddlers, Rev	Caregiver Rating	x			Hybrid	10 minutes	
CBCL/ABC	Caregiver Rating	x	x	x	Hybrid	15 minutes	
BRIEF P/2/A	Caregiver Rating	X (2.0)	x	x	Hybrid	15 minutes	
Communication and Symbolic Behavior Scale	Direct assessment	x			In-person	30 minutes	
Beery VMI	Direct assessment	X (2.0)	x	x	In-person	15 minutes	
PPVT-4/PPVT-3 Spanish	Direct assessment	x (2.5)	x	x	In-person	15 minutes	

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Minimal



Measure	Type	Age Group			In-Person/ Remote/ Hybrid	Estimated Time to Complete	Cadence (One time, annually, etc.)
		0-5	6-21	22+			
Cat Dog Stroop	Direct assessment		x	x	In-person	5 minutes	
Down syndrome Mental Status Exam	Direct assessment			x	In-person	30 minutes	
TBD (Dev/Cog)	Direct assessment	x	x	x	In-person	45-60 minutes	
DAS-2 Digits and Picture Rec	Direct assessment		x	x	In-person	10 minutes	
TBD (processing speed)	Direct assessment		x	x	In-person	5 minutes	
Verbal Fluency	Direct assessment		x	x	In-person	5 minutes	

	0-5	6 to 21	22+	
Caregiver		110	75	30
Direct		120	115	145


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Consent WG update

Will likely model off other large network multi-site/institution studies (such as ECHO). Will be using some remote delivery (e.g., redcap) and will include graphics and better visuals to explain activities. Consents will have compliant signature (e.g., docusign, vera). Single IRB model (institution or company to house this still TBD). Will also provide supporting materials (social stories, videos) and retention tools (e.g., newsletters, swag). Getting input and ideas from CAB. Goal for IRB creation late spring following approval of core protocol.

Remote Digital Health WG - Minimal Core Protocol

Both sleep measures will not pertain to IBIS (3+ or 12+) but will have remote option. Collecting both will also help validation for the Nox (which is needed for DS). Actigraph will also be 3+ so not going to be part of IBIS data collection. Will continue to discuss adding sleep questionnaire, maybe also something about diet. Overall three slides for minimal (2 sleep, 1 activity):


Minimal 

Data Collection Measure	Age Group	In-Person/Remote/Hybrid	Estimated Time to Complete	Cadence (One time, annually, etc.)
WatchPAT (Sleep)	FDA approved for 12 years of age and up.	Remote set-up and use.	1 night	Every 2 years

Additional Information

- User friendly and minimally invasive.
- Central reading center TBD.

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Minimal 


Data Collection Measure	Age Group	In-Person/Remote/Hybrid	Estimated Time to Complete	Cadence (One time, annually, etc.)
Nox Device (Sleep)	To be utilized in the pediatric population (3 years of age and older)	Requires a technician to set up and can be used remotely or in-person	1 night	Every 2 years

Additional Information

- Requires a validation group between NOX and WatchPAT of 30-40 individuals dependent upon budget.
- In-person assessments could potentially be conducted at select sites such as Saint Jude's and Texas Children's Hospital.
- Central reading center TBD.

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Minimal



Data Collection Measure	Age Group	In-Person/ Remote/ Hybrid	Estimated Time to Complete	Cadence (One time, annually, etc.)
ActiGraph LEAP (Activity and Sleep)	To be utilized in participants 3 years of age and older	Remote or hybrid set-up and use	7 days	Every 12 months


Additional Information

- There are no validated PA cut points for young children in this population.
- Sample rate of 32 Hz for activity data, which would provide 32 days of battery life and 79 days of data storage.
- Staggering data collection periods could help account for seasonal variations in activity level.

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We could opt to collect some activity data from the wish list....(leg band).

Wish List



Data Collection Measure	Age Group	In-Person/ Remote/ Hybrid	Estimated Time to Complete	Cadence (One time, annually, etc.)
Activity Leg Band (Activity)	To be utilized in participants 1-3 years of age	Remote or hybrid set-up and use	7 days	Every 12 months
Activity (Activity)	To be utilized in children/adults who cannot tolerate ActiGraph	Remote or hybrid set-up and use	7 days	Every 12 months
Parent/Caregiver Activity Data	To be utilized in parents/caregivers of children participants	Remote or hybrid set-up and use	7 days	Every 12 months when child/loved one completes data collection

Additional Information

- Staggering data collection periods could help account for seasonal variations in activity level.

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DS-CDP TIMELINE:

Feb-May - WG will continue meeting, likely on bimonthly schedule

May 2025 – WG present final protocol for DS-CD to steering committee for review/approval

June 2025 – Final common protocol and supporting docs submitted to IB