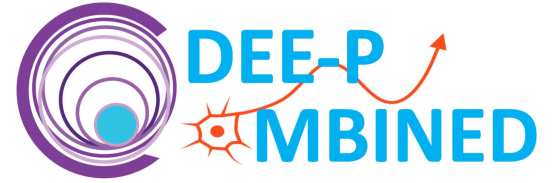


OTHER PROMISING MEASURES



Patient-Reported Outcome Measures

ORCA - Observer Reported Communication Measure

Event-Related Potentials

Audio - Language processing

Visual - Facial recognition, Object recognition

Home-based video measures

Seizure-detecting glasses - Eysz

Facial recognition software - PainChek

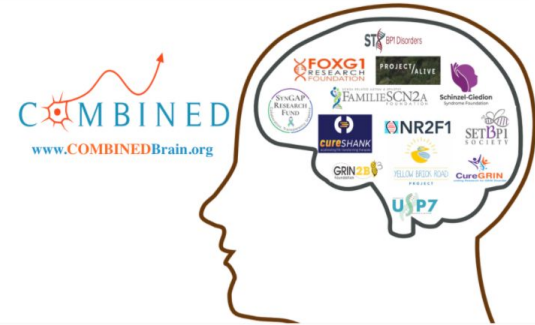
Video-based motor measures - Casimir, Actimyo

COMBINEDBrain - FDA Grant

Expanding the Observer-Reported Communication Ability (ORCA) Measure: Measuring the Communication Ability of Individuals With Rare, Neurodevelopmental Disorders



Observer-Reported Communication Ability Measure



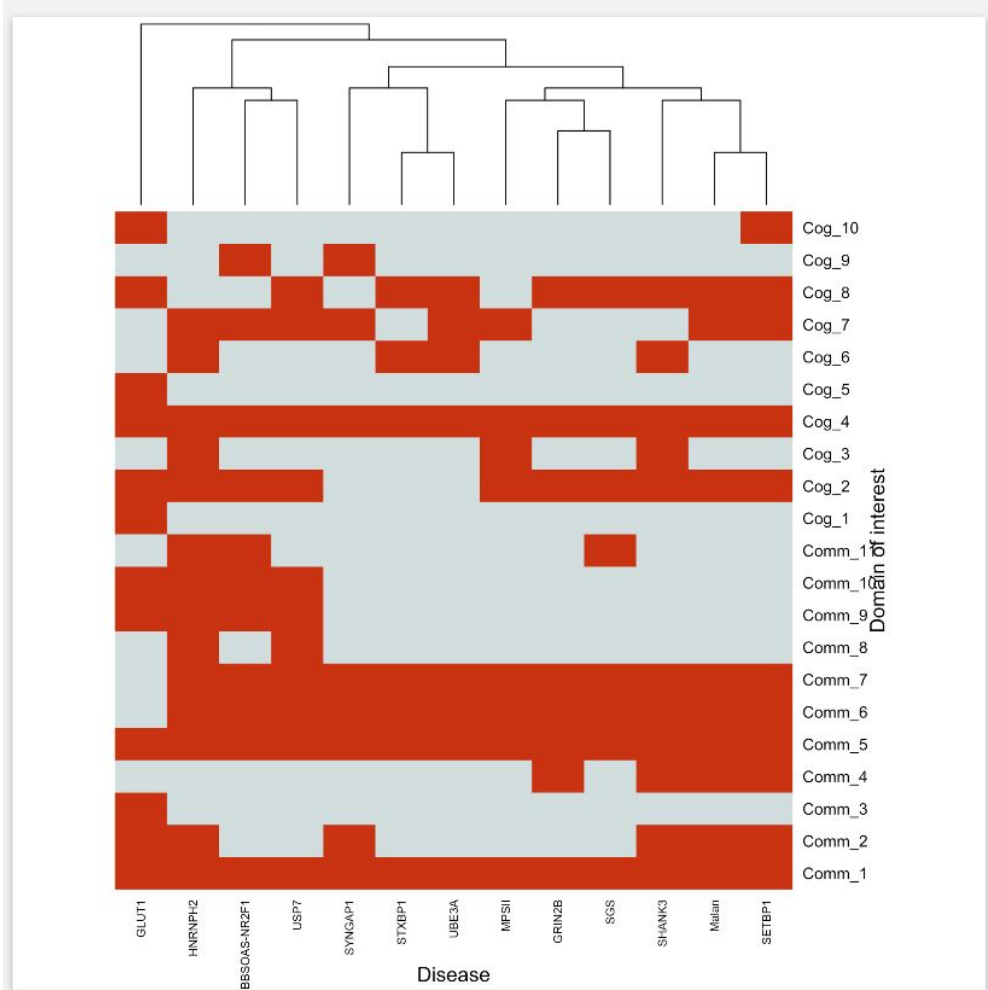
When assessing the communication ability of individuals with neurodevelopmental disorders (NDDs) in the context of clinical trials, existing clinical outcome assessments (COAs) have significant limitations, including the inability to differentiate among individuals and lack of input from parent advocates.

With a grant from the U.S. Food and Drug Administration (FDA), Dr. Christy Zigler (Principal Investigator) and other CHM investigators will work with COMBINEDBrain, a patient advocacy consortium, to expand the ORCA measure to a range of NDD populations.



Observer-Reported Communication Ability Measure

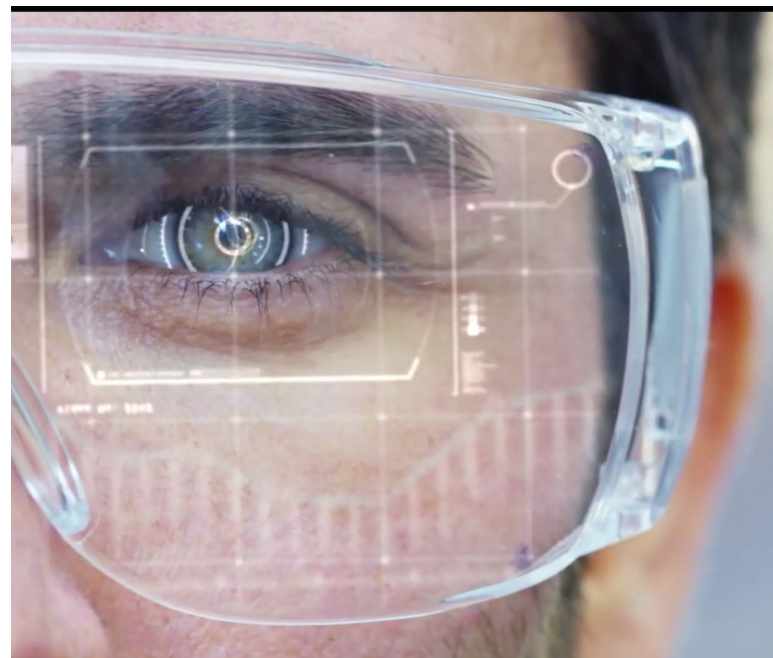
- **Angelman-like:**
 - SynGAP1
 - STXBP1
- **Moderately Angelman-like**
 - Phelan-McDermid - SHANK3
 - Hunter - MPSII
 - Malan - NFIX
 - SETBP1 deficiency
 - SCN2A
 - GRIN
- **Not-Angelman-like**
 - HNRNPH2
 - Hao-Fountain - USP7
 - BBSOAS - NR2F1
- **Very severely affected**
 - Schinzel-Giedion - SETBP1 over-expression



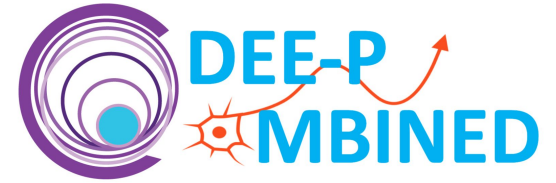
EYSZLAB.com



TRANSFORMING EPILEPSY CARE



PAINCHEK



Artificial intelligence

Facial recognition software

BMC Geriatrics

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Research article | [Open Access](#) | [Published: 28 May 2021](#)

Evaluation of the Psychometric Properties of PainChek® in UK Aged Care Residents with advanced dementia

[Ivana Babicova](#) , [Ainslea Cross](#), [Dawn Forman](#), [Jeffery Hughes](#) & [Kreshnik Hoti](#)

[BMC Geriatrics](#) **21**, Article number: 337 (2021) | [Cite this article](#)

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HOME-BASED VIDEO OUTCOMES



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Turning Patient Voices into Better Data

[learn more](#)



INTEGREVIEW

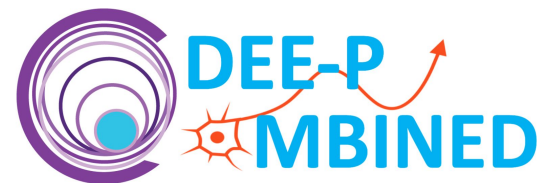
November 17, 2023

APPROVED

Angelman Syndrome- Remote Video Capture Study

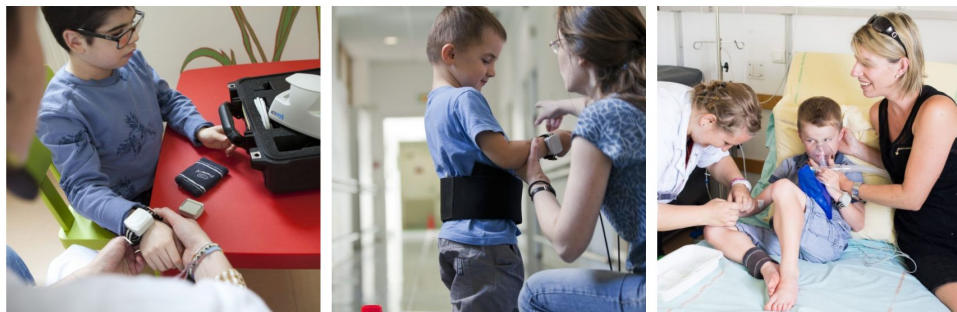
Casimir, a clinical research organization specializing in incorporating patient-reported data into clinical trials, Boston Children's Hospital, and Ionis Pharmaceuticals are in the process of developing an at-home assessment for potential use in clinical trials that would capture patients with Angelman syndrome (AS) in their everyday environment. To date, the outcome measures used in clinical trials for AS patients have focused mostly on in-clinic assessments.

HOME-BASED VIDEO OUTCOMES



ActiMyo

ActiMyo is a new tool developed to evaluate the physical condition of subjects suffering from pathologies associated with movement disorders. It is the result of close collaboration between the [Institute of Myology](#) and [Sysnav](#), an innovative French company, specialized in navigation systems and motion analysis. It was developed to record in a very precise and sensitive way, the movements of daily life of a patient in his/her usual environment.



Chabaron (2018) Prospective and longitudinal natural history study of patients with Type 2 and 3 spinal muscular atrophy: Baseline data NatHis-SMA study

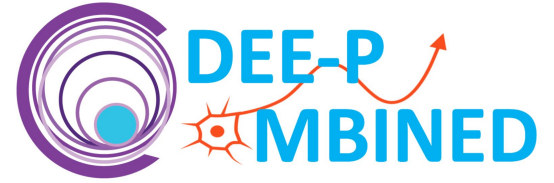
STRENGTH	Pulmonary Function Tests	FVC	0.780	
		MEP	0.824	
		MIP	0.509	
		PCF	0.701	
		SNIP	0.331	
	Myogrip	Dominant side	0.876	
		Non-dominant side	0.921	
	Myopinch	Dominant side	0.907	
		Non-dominant side	0.928	
	Timed Tests	6MWT	0.818	
		10MWT	-0.721	
		Rise from floor	-0.738	
		Climb 4 stairs	-0.830	
Descend 4 stairs		-0.770		
UPPER LIMBS FUNCTION	ActiMyo	Activity time	0.694	
		nGyr	0.714	
		nAcc	0.879	
		zAcc	0.847	
		Power	0.515	
	Moviplate	Dominant side	0.719	
		Non-dominant side	0.690	
	Active-Seated		Functional Reaching Volume	0.899
	Electrophysiology	Ulnar-ADM	1st CMAP response Amplitude	0.484
		Accessory-trapezius		0.551
		Radial-anconeus		0.690
		Peroneal-tibialis anterior		0.770
	Arm	Biceps	C-CSA	0.541
			Fat fraction	-0.845
		Triceps	C-CSA	0.603
			Fat fraction	-0.740
	Flexor	C-CSA	Fat fraction	0.551
				0.551

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Fig 4. Correlation matrix between motor function (MFM) total score and other outcome measures. Abbreviations: FVC: Forced Vital Capacity; MEP: Maximum Expiratory Pressure;

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Fig 4. Correlation matrix between motor function (MFM) total score and other outcome measures. Abbreviations: FVC: Forced Vital Capacity; MEP: Maximum Expiratory Pressure;

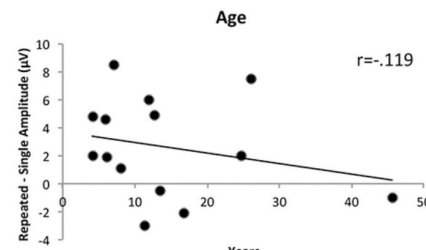
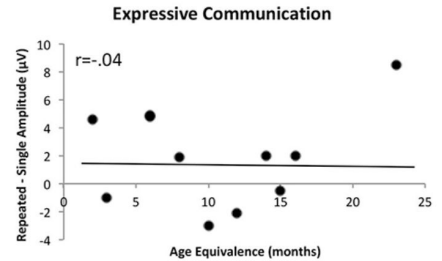
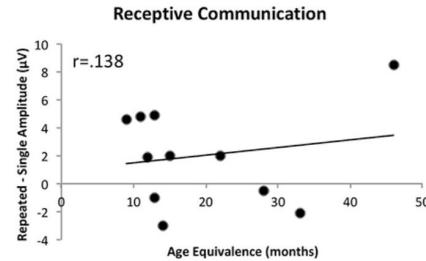
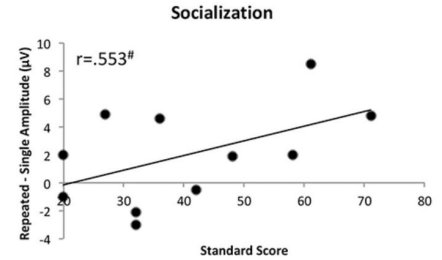
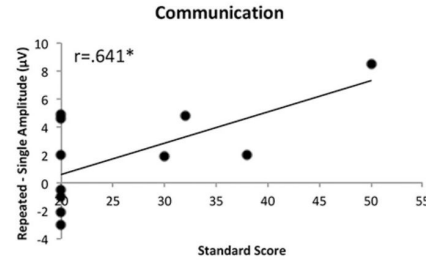
EVENT-RELATED POTENTIALS



ERP/EP
Event Related Potentials/
Evoked Potentials



Key (2018), Feasibility of using auditory event-related potentials to investigate learning and memory in nonverbal individuals with Angelman syndrome



EVENT-RELATED POTENTIALS

INVITED REVIEW

Searching for a “Brain Signature” of Neurodevelopmental Disorders: Event-Related Potentials and the Quest for Biomarkers of Cognition

Alexandra P. Key

Vanderbilt University Medical Center, Vanderbilt Kennedy Center, Nashville, Tennessee, U.S.A.

- Avoids floor effect
 - Masked data acquisition
 - Passive paradigm
 - Changes in brain activity before behavioral symptoms
-
- Visual Face recognition
 - Auditory language processing

Carson (2021), Nutritional Formulation for Patients with Angelman Syndrome: A Randomized, Double-Blind, Placebo-Controlled Study of Exogenous Ketones

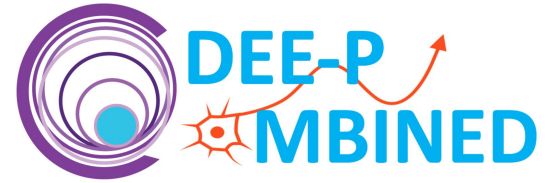
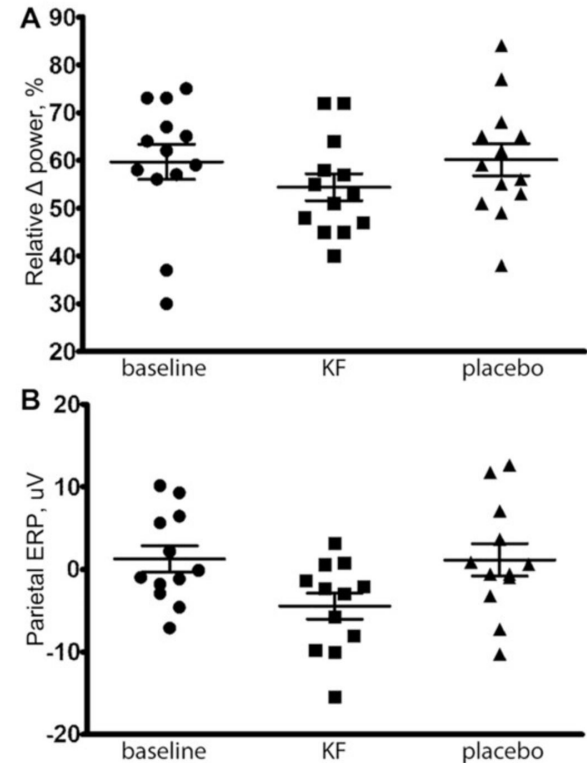
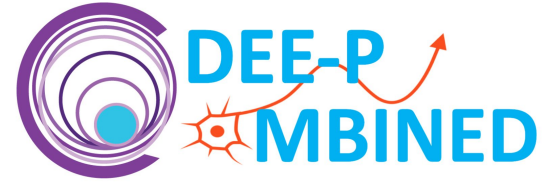


FIGURE 3



SLEEP: The Final Frontier



- Polysomnography
- Actigraphy
- Videography
- Portable devices
 - Infrared
 - Bed Mat
- Patient-Reported Surveys
 - Children's Sleep Habits Questionnaire (CSHQ)
 - Parents' Sleep Habits Questionnaire
 - Excessive Daytime Sleepiness
- Circadian measures
 - Heart rate
 - Temperature

