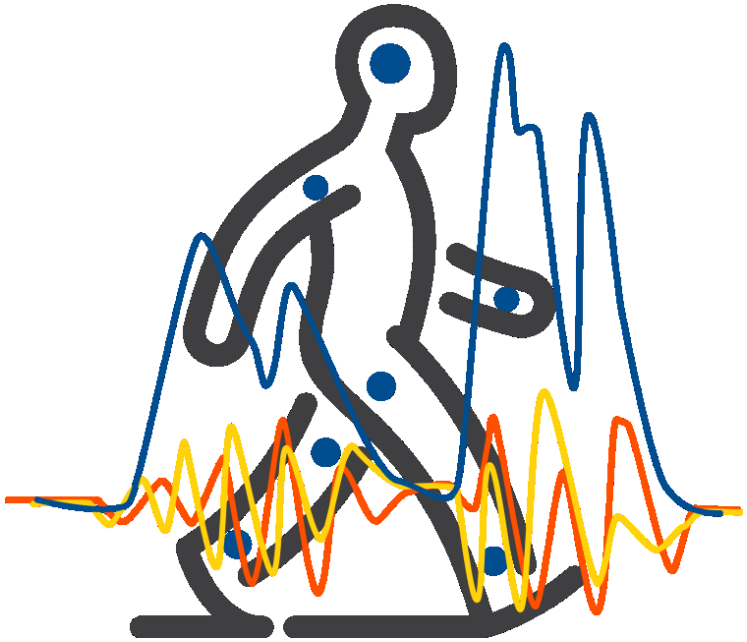
European Reference Network
for rare or low prevalence complex diseases
Network Neurological Diseases (ERN-RND)

ean european academy of neurology

European Reference Network
for rare or low prevalence complex diseases
Network Neuromuscular Diseases (ERN EURO-NMD)

DG ‚Ataxia and HSP'
24. November 2020

Joint webinar series

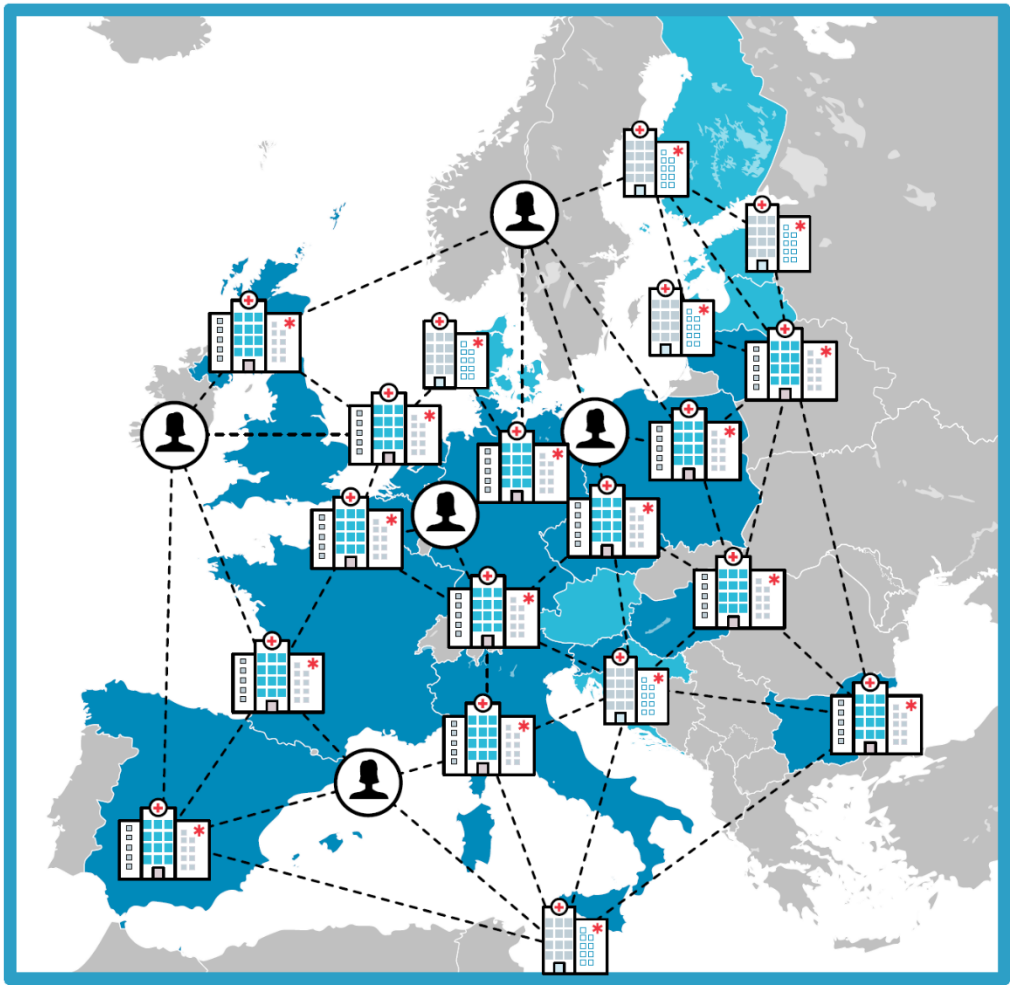


'Development of SARA@home: a novel assessment tool for patients with ataxia'

by Gessica Vasco & Susanna Summa
Ospedale Pediatrico Bambino Gesù, Rome, Italy

European Reference Network for RARE Neurological Diseases (ERN-RND)

- **Countries with Full Members**
- **Countries with Affiliated Partners**



ERN-RND covers 6 disease groups:

1. Ataxia and HSP
2. Leukodystrophies
3. Dystonias /NBIA/Paroxysmal disorders
4. Chorea and HD
5. FTD
6. Atypical Parkinsonism

General information about the webinars

- Focus on : RARE neurological, neuromuscular and movement disorders and **neurorehabilitation**
- 40-45min presentation
- 15min Q&A session at the end (please write your questions in the Q&A)
- Recorded Webinar and presentation to be found at the latest 2 weeks after on: http://www.ern-rnd.eu/education-training/past-webinars/
- Further information: http://www.ern-rnd.eu/disease-knowledge-hub/ataxia/
- Post-webinar survey (2-3min): satisfaction, topic/speaker ideas for next webinars

ePAG: european Patient Advocacy Groups

Mary Kearney

Friedreich's Ataxia Research Alliance Ireland (FARA)

In ERN-RND Patient Advocate for: **Ataxia/HSP**

Speakers: Gessica Vasco and Susanna Summa

Gessica Vasco

- MD, PhD in Pediatric Neurology at Catholic University of Rome
- Since 2013 Pediatric neurologist at Bambino Gesù Children Hospital, Neurorehabilitation center Rome
- Research focus: neurodegenerative and neuromuscolar disorders, such as Friedreich ataxia
- Early Onset Ataxia, Duchenne Muscular Dystrophy, ranging from bench work to clinical studies.
- Member of the natural history European consortia EFACTS for FA

Contact: gessica.vasco@opbg.net

Susanna Summa

Training: PhD in Bioengineering at University of Genoa
Current position: Research contract at the Bambino Gesù Children Hospital
Research focus: Movement analysis of pediatric patients with ataxia and human-machine interaction for the assessment of neuromotor diseases and for neuromotor recovery with robotic platforms.

Contact: susanna.summa@opbg.net

Webinar outline

- Introduction
- Cerebellar ataxia: Clinical examination
- Ataxias Rating Scales
- Research project: Pediatric ataxias and Public Health
- Digital assessment tools
- The Sara@home
- Conclusion and key points

Q1: What is your professional background?

a) Neurologist

b) Neuropediatrician

c) Physiatrist

d) Geneticist

e) Nurse

f) Physiotherapist

g) Speech therapist

h) Occupational therapist

i) Biomedical engineer

Ataxia

Disorganized, poorly coordinated or clumsy movement

Classification:

Non Genetic

- Acute (acquired)
- Recurrent

Genetic

- Progressive
- Non progressive- congenital

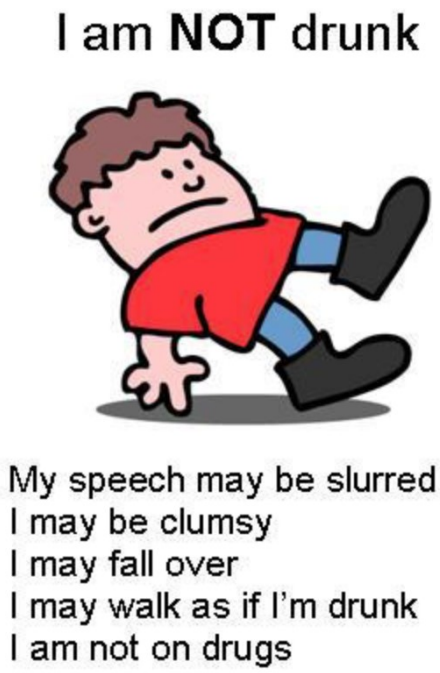
Different types:

- Cerebellar
- Proprioceptive
- Vestibular

Cerebellar ataxia: clinical examination

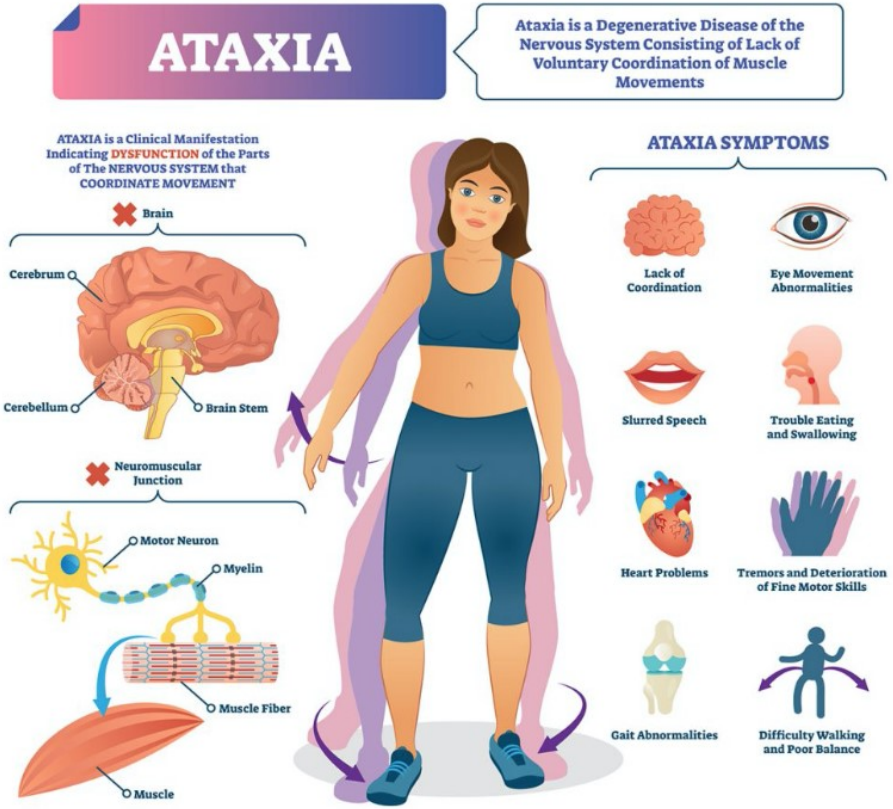
- GAIT
  - Look normal gait, including turns
  - Wide-based, problems with tandem walking, steps are variable
- LIMB
  - Look for kinetic tremor and for dysmetria

    Finger Nose and Heel Shin
- STANCE
  - Normal and tandem stance
- TRUNK
  - Sitting without support



Cerebellar ataxia: clinical examination

- EYE MOVEMENTS
  - fixation: instability, square wave jerks
  - pursuit gaze evoked nystagmus
  - saccades jerky, interrupted pursuit
- SPEECH
  - Spontaneus speech, PATA repetition



Don‘t forget…..

Early signs

- Infantile hypotonia
- Motor delay
- Speech delay
- Ocular dyspraxia / nystagmus
- Seizure

Non-cerebellar manifestations

- Spasticity
- Peripheral neuropathy / afferent deficits
- Parkinsonism
- Dystonia
- Myoclonus

How can I measure ataxia?

RATING SCALES

- **BARS** Brief Ataxia Rating Scale
- **FAIS** Friedreich ataxia impact scale
- **FARS** Friedreich's Ataxia Rating Scale
- **ICARS** International cooperative ataxia rating scale
- **MICARS** Modified ICARS
- **DSI-ARSACS** Disease severity Index for ARSACS
- **NESSCA** Neurological Examination Score for spinocerebellar Ataxia
- **FXTAS-RS** Fragile X associated Tremor Ataxia Syndrome Rating Scale
- **UMSARS** unified multiple system atrophy rating scale
- **SARA** Scale for the Assessment and Rating of Ataxia
- **INAS** Inventory of Non-Ataxia Signs

FUNCTIONAL TEST

- **AFCS** Ataxia Functional composite scale
- **APP-Coo- Test**
- **SCAFI** Spinocerebellar ataxia Functional Index
- **HEVELIUS**
- **CCFS** Composite Cerebellar Functional Severity Score

Q2: Which is the major challenge in assessing ataxia?

a) Acceptability of assessment

b) Overcoming ceiling and floor effects

c) Validation for all cerebellar disorders

d) Inter-raters reproducibility

e) Realiability for children younger then 12

f) Continuous and remote monitoring

MDS COMMISSIONED REVIEWS

Assessment of Ataxia Rating Scales and Cerebellar Functional Tests: Critique and Recommendations

Santiago Perez-Lloret, MD, PhD,[1,2,3*] Bart van de Warrenburg, MD, PhD,[4] Malco Rossi, MD, PhD,[5] Carmen Rodríguez-Blázquez, PhD, MSc,[6] Theresa Zesiewicz, MD, FAAN,[7] Jonas A.M. Saute, MD, PhD,[8,9,10,11] Alexandra Durr, MD, PhD,[12] Masatoyo Nishizawa, MD, PhD,[13] Pablo Martinez-Martin, MD, PhD,[14] Glenn T. Stebbins, PhD,[15] Anette Schrag, MD, PhD,[16] and Matej Skorvanek, MD, PhD,[17,18] and members of the MDS Rating Scales Review Committee

SARA and ICARS most used scales in the literature

ICARS good responsivness in SCA e FA
SARA better dimensionality and better reproducibility
Shortness

Movement Disorders, 2020

How can I measure ataxia?

RATING SCALES

- **BARS** Brief Ataxia Rating Scale
- **FAIS** Friedreich ataxia impact scale
- **FARS** Friedreich's Ataxia Rating Scale
- **ICARS** International cooperative ataxia rating scale
- **MICARS** Modified ICARS
- **DSI-ARSACS** Disease severity Index for ARSACS
- **NESSCA** Neurological Examination Score for spinocerebellar Ataxia
- **FXTAS-RS** Fragile X associated Tremor Ataxia Syndrome Rating Scale
- UMSARS unified multiple system atrophy rating scale
- **SARA** Scale for the Assessment and Rating of Ataxia
- **INAS** Inventory of Non-Ataxia Signs

FUNCTIONAL TEST

- **AFCS**Ataxia Functional composite scale
- **APP-Coo-test**
- **SCAFI** Spinocerebellar ataxia Functional Index
- **HEVELIUS**
- **CCFS** Composite Cerebellar Functional Severity Score

CLINIMETRIC PROPERTIES

STRENGHTS

WEAKNESSES

SARA and ICARS most used scales in the literature

ICARS good responsivness in SCA e FA

SARA better dimensionality and better reproducibility

Shortness

LIMITATIONS

Ceiling effects

Floor effects

Lack of validation for another cerebellar disorders

Cerebellar assessment in children

- Fatigue testing
- Developmental delay/intellectual disability
- Age-related maturation of the nervous system is associated with improved coordination and fine motor skills.
- Age validation

Published in final edited form as:
Neuroimage. 2010 January 1; 49(1): 63–70. doi:10.1016/j.neuroimage.2009.08.016.

Cerebellum development during childhood and adolescence: a longitudinal morphometric MRI study

Henning Tiemeier[1,2], **Rhoshel K. Lenroot**[1], **Deanna K. Greenstein**[1], **Lan Tran**[1], **Ronald Pierson**[3], and **Jay N. Giedd**[1]

Brain and Behavior

Open Access

The developing human brain: age-related changes in cortical, subcortical, and cerebellar anatomy

Dafna Sussman[1], Rachel C. Leung[2], M. Mallar Chakravarty[3,4], Jason P. Lerch[5,6] & Margot J. Taylor[2]

0031-3998/11/6901-0080
PEDIATRIC RESEARCH

Vol. 69, No. 1, 2011
Printed in U.S.A.

The Cerebellar Development in Chinese Children—A Study by Voxel-Based Volume Measurement of Reconstructed 3D MRI Scan

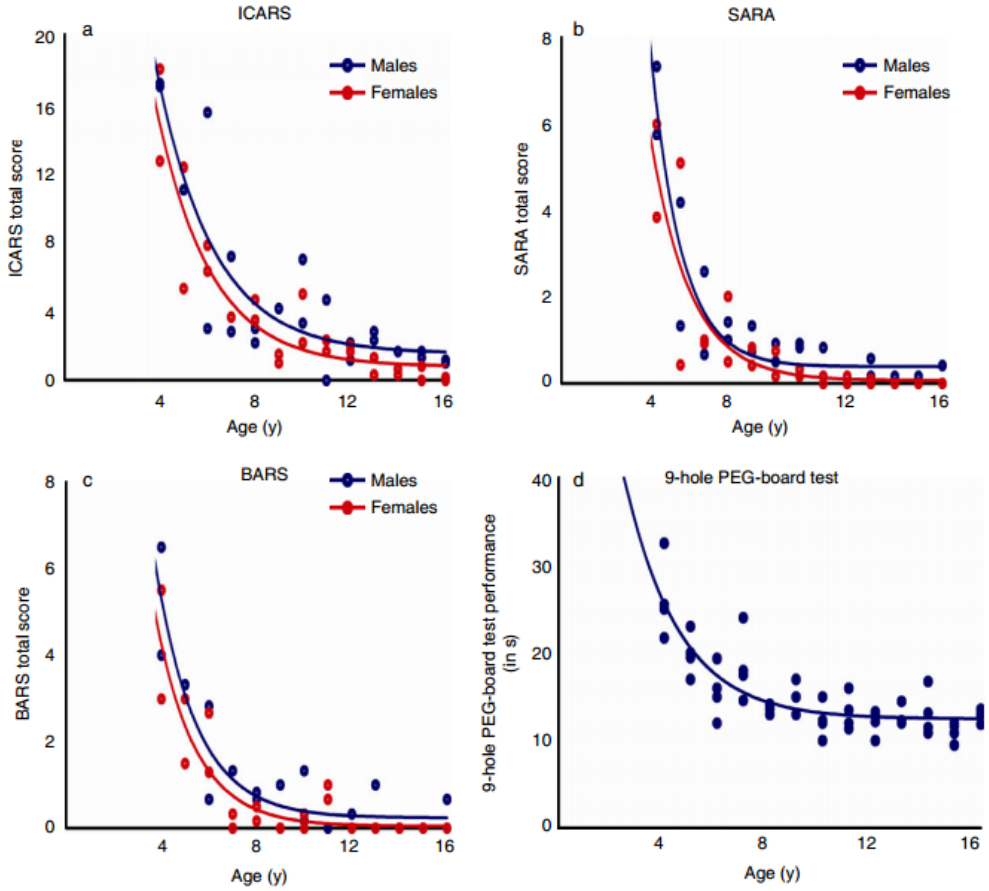
KUAN-HSUN WU, CHIA-YUAN CHEN, AND EIN-YIAO SHEN

Departments of Pediatrics [K.-H.W.] and Radiology [C.-Y.C.], Taipei Medical University, Wan Fang Hospital, Taipei 116, Taiwan; Graduate Institute of Acupuncture Science [E.-Y.S.], China Medical University, Taichung 404, Taiwan; Department of Pediatrics [E.-Y.S.], China Medical University Hospital, Taipei Branch, Taipei 114, Taiwan

DEVELOPMENTAL MEDICINE & CHILD NEUROLOGY ORIGINAL ARTICLE

Ataxia rating scales are age-dependent in healthy children

RICK BRANDSMA[1*] | ANNE H SPITS[1*] | MARIEKE J KUIPER[1] | ROELINKA J LUNSING[1] | HUIBERT BURGER[2] | HUBERTUS P KREMER[1] | DEBORAH A SIVAL[3] | ON BEHALF OF THE CHILDHOOD ATAXIA AND CEREBELLAR GROUP[†]



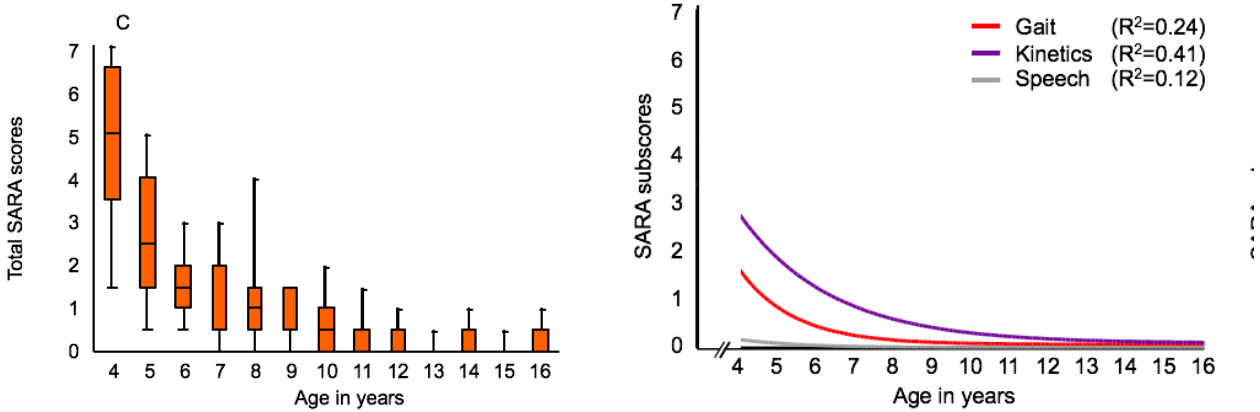
Ataxia rating scales (ICARS, SARA and BARS) and the 9-hole PEG-board test related to age

Population 52 healthy children
Age 4-16 years

Results
ICARS, SARA, BARS and PEG-board test outcomes were age-dependent
ICARS 12,5 yr
SARA 10 yr
BARS 11 yr
9 HPT 11,5 yr

Age-related reference values for the pediatric Scale for Assessment and Rating of Ataxia: a multicentre study

TJITSKE F LAWERMAN[1]* | RICK BRANDSMA[1]* | HUIBERT BURGER[2] | JOHANNES G M BURGERHOF[3] | DEBORAH A SIVAL[4] | ON BEHALF OF THE CHILDHOOD ATAXIA AND CEREBELLAR GROUP OF THE EUROPEAN PEDIATRIC NEUROLOGY SOCIETY[†]



Results: SARA scores were related with age (r=-0.779, p<0.001). Age explained 47% of SARA scores (R2=0.47). The youngest children revealed the highest scores and the highest variation in scores (≤ 7 years; p<0.001). After 12 years of age, pediatric scores approached adult outcomes. Inter-observer agreement (Intraclass Correlation Coefficient: 0.69) revealed a positive relationship with age (p<0.001).

SARA (Scale for the assessment and rating of ataxia)

Total score: 40

1. Gait (score 0-8)
2. Stance (score: 0-6)
3. Sitting (score: 0-4)
4. Speech disturbance (score: 0-6)
5. Finger chase (score: 0-4)
6. Nose-finger test (score: 0-4)
7. Fast alternating hand movements(score: 0-4)
8. Heel-shin slide (score: 0-4)

1) Gait	2) Stance
Proband is asked (1) to walk at a safe distance parallel to a wall including a half-turn (turn around to face the opposite direction of gait) and (2) to walk in tandem (heels to toes) without support. 0 Normal, no difficulties in walking, turning and walking tandem (up to one misstep allowed) 1 Slight difficulties, only visible when walking 10 consecutive steps in tandem 2 Clearly abnormal, tandem walking >10 steps not possible 3 Considerable staggering, difficulties in half-turn, but without support 4 Marked staggering, intermittent support of the wall required 5 Severe staggering, permanent support of one stick or light support by one arm required 6 Walking > 10 m only with strong support (two special sticks or stroller or accompanying person) 7 Walking < 10 m only with strong support (two special sticks or stroller or accompanying person) 8 Unable to walk, even supported	Proband is asked to stand (1) in natural position, (2) with feet together in parallel (big toes touching each other) and (3) in tandem (both feet on one line, no space between heel and toe). Proband does not wear shoes, eyes are open. For each condition, three trials are allowed. Best trial is rated. 0 Normal, able to stand in tandem for > 10 s 1 Able to stand with feet together without sway, but not in tandem for > 10s 2 Able to stand with feet together for > 10 s, but only with sway 3 Able to stand for > 10 s without support in natural position, but not with feet together 4 Able to stand for >10 s in natural position only with intermittent support 5 Able to stand >10 s in natural position only with constant support of one arm 6 Unable to stand for >10 s even with constant support of one arm
Score	Score
3) Sitting	**4) Speech disturbance**
Proband is asked to sit on an examination bed without support of feet, eyes open and arms outstretched to the front. 0 Normal, no difficulties sitting >10 sec 1 Slight difficulties, intermittent sway 2 Constant sway, but able to sit > 10 s without support 3 Able to sit for > 10 s only with intermittent support 4 Unable to sit for >10 s without continuous support	Speech is assessed during normal conversation. 0 Normal 1 Suggestion of speech disturbance 2 Impaired speech, but easy to understand 3 Occasional words difficult to understand 4 Many words difficult to understand 5 Only single words understandable 6 Speech unintelligible / anarthria
Score	Score

SARA (Scale for the assessment and rating of ataxia)

ADVANTAGES

- Simple administration
- Mean time to administer SARA in patients was 14.2±7.5 minutes
- Inter-intra rater reliability is high

DISADVANTAGES

- Not usable for the diagnosis of the disease in the initial phase
- SARA is reliably applicable to children beyond the age of **12** years
- Low sensitivity

"Scale for the assessment and rating of ataxia: development of a new clinical scale."
Schmitz-Hubsch, et al.

ELSEVIER

Neurologic Clinics

Volume 38, Issue 2, May 2020, Pages 231-254

Clinical Rating Scales and Quantitative Assessments of Movement Disorders

Arjun Tarakad MD ✉

Department of Neurology, Parkinson's Disease Center and Movement Disorders Clinic, Baylor College of Medicine, 7200 Cambridge Street Suite 9A, Houston, TX 77030, USA

- A more recent and fast developing area within the assessment of movement disorders is the use of computer-assisted technologies in quantifying disease characteristics.
- Two categories:
  - Wearable devices
  - Digital interfaces

Pediatric ataxias and Public Health : epidemiological studies and disease registry, characterization of genetic determinants and implementation of protocols for diagnosis, management, and rehabilitation using innovative low cost, widely accessible technologies (NET-2013-02356160)

2016/17 → **2019/20**

Ministero della Salute

WP1
Istituto Superiore di Sanità
PI N. Vanacore
Pediatric ataxias in Italy: epidemiological studies and disease registry, development of a multilevel informatic platform for clinicians and families, and implementation of guidelines for diagnosis, management and care within the National Health System

WP4
IRCCS Eugenio Medea
PI R. Borgatti
Cognitive and behavioral defects in non-progressive pediatric ataxias: systematic cognitive profiling, innovative neuroimaging studies and development of low-cost, widely accessible technologies for personalized home-based rehabilitation

WP2
IRCCS Fondazione Santa Lucia
PI E.M. Valente
Improving genetic diagnosis of pediatric ataxias: identification of novel genetic determinants, large scale molecular screenings and genotype-phenotype correlates by use of state-of-the-art genomic technologies

WP3
IRCCS O. P. Bambino Gesù
PI E. Bertini
Development of innovative low-cost, widely accessible technologies for quantitative assessment and home-based rehabilitation of motor function in pediatric ataxias

Home-based Monitoring

WP3

IRCCS O. P. Bambino Gesù

PI E. Bertini

Development of innovative low-cost, widely accessible technologies for quantitative assessment and home-based rehabilitation of motor function in pediatric ataxias

SARA@home

Institute of Clinical Physiology (IFC-CNR Messina)

Rehab@home

By 2D virtual reality («serious games») and IMU control

Q3: Which are the benefits of technologies for the remote assessment?

- Low-cost
- Sharing data/informations
- Continuous monitoring
- Objective assessment
- Patients Acceptance
- All the previous

SARA@home Objectives

- To digitalize the SARA scale
- Continuous monitoring
- To avoid uncomfortable situations during evaluation

Computer Methods and Programs in Biomedicine
Volume 188, May 2020, 105257

Development of SaraHome: A novel, well-accepted, technology-based assessment tool for patients with ataxia

Susanna Summa [a], Tommaso Schirinzi [a, b], Giuseppe Massimo Bernava [c], Alberto Romano [a], Martina Favetta [a], Enza Maria Valente [d, e], Enrico Bertini [f], Enrico Castelli [a], Maurizio Petrarca [a], Giovanni Pioggia [c, 1], Gessica Vasco [a, 1]

QUEST	Subscales		Total score
	Assistive device	Services	
very positive feedback	6	9	9
positive feedback	4	1	1
negative feedback	0	0	0
very negative feedback	0	0	0

- High interest and participation from patients.
- Substantial satisfaction and a perception of ease of use from the parents involved in the assessment.

IMI	Subscales					
	Interest/Enjoyment	Perceived Competence	Effort/Importance	Pressure/Tension	Perceived Choice	Value/Usefulness
positive feedback	6	7	10	10	6	9
negative feedback	3	3	0	0	4	1
neutral feedback	1	0	0	0	0	0

Q4: Which items drives SARA progression?

- Gait
- Stance
- Sitting
- Speech disturbance
- Finger chase
- Nose-finger test
- Fast alternating hand movements
- Heel-shin slide

Neurology Genetics

A peer-reviewed clinical and translational neurology open access journal

Home Articles Issues

June 2020; 6 (3) ARTICLE
OPEN ACCESS

Neurologic outcomes in Friedreich ataxia

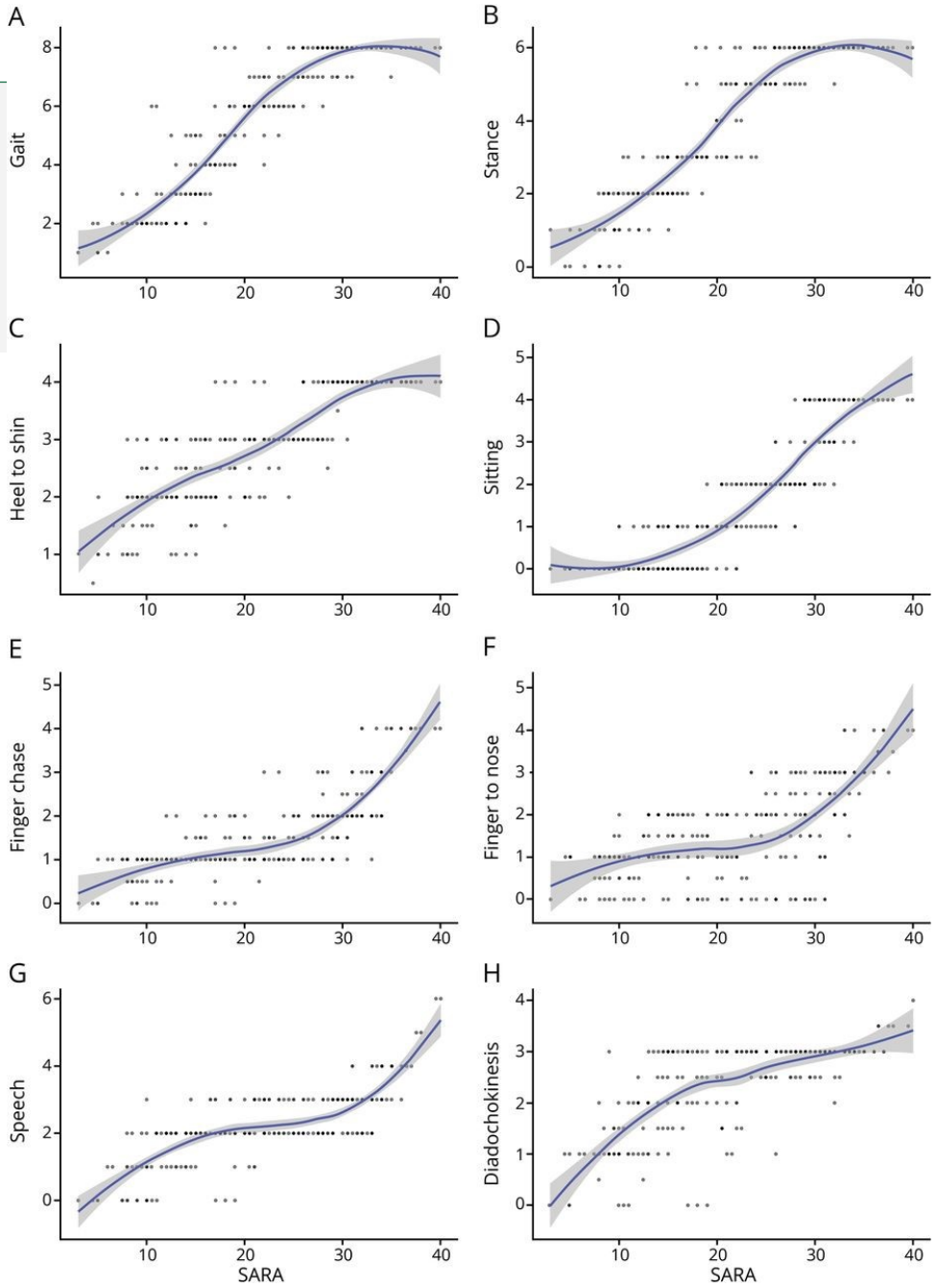
Study of a single-site cohort

Massimo Pandolfo

First published March 20, 2020, DOI: https://doi.org/10.1212/NXG.0000000000000415

The present study analyzes a single EFACTS site cohort of 54 patients with FRDA with the aim of characterizing the pattern of disease progression and identifying the most rapidly progressing subset of patients.

the European Friedreich's Ataxia Consortium for Translational Studies (EFACTS)



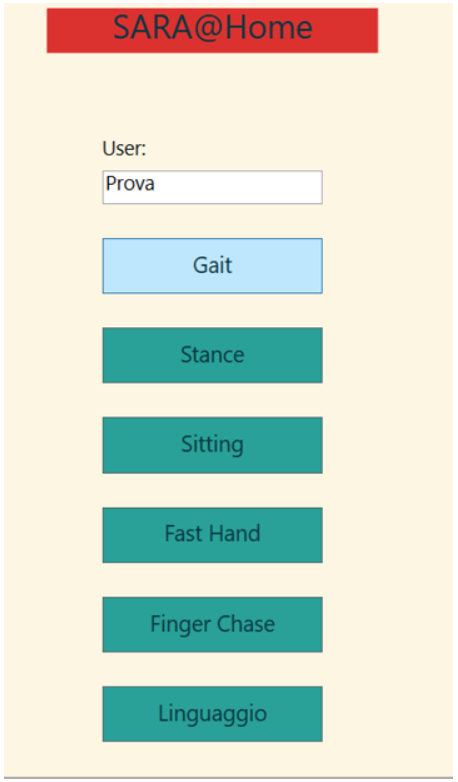
SARA@home: digital interface

1. Gait (score 0-8) ✓
2. Stance (score: 0-6) ✓
3. Sitting (score: 0-4) ✓
4. Speech disturbance (score: 0-6)

   Evaluated through PATAtest ✓
5. Finger chase (score: 0-4) ✓
6. Nose-finger test (score: 0-4) ✗
7. Fast alternating hand

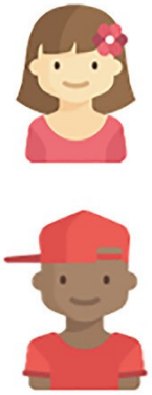
   movements(score: 0-4) ✓
8. Heel-shin slide (score: 0-4) ✗

Software



SARA@Home App

2) Collect data



1) Generate user interface

Restful interfac

Proteo

Apollo

Bifrost PLUGIN

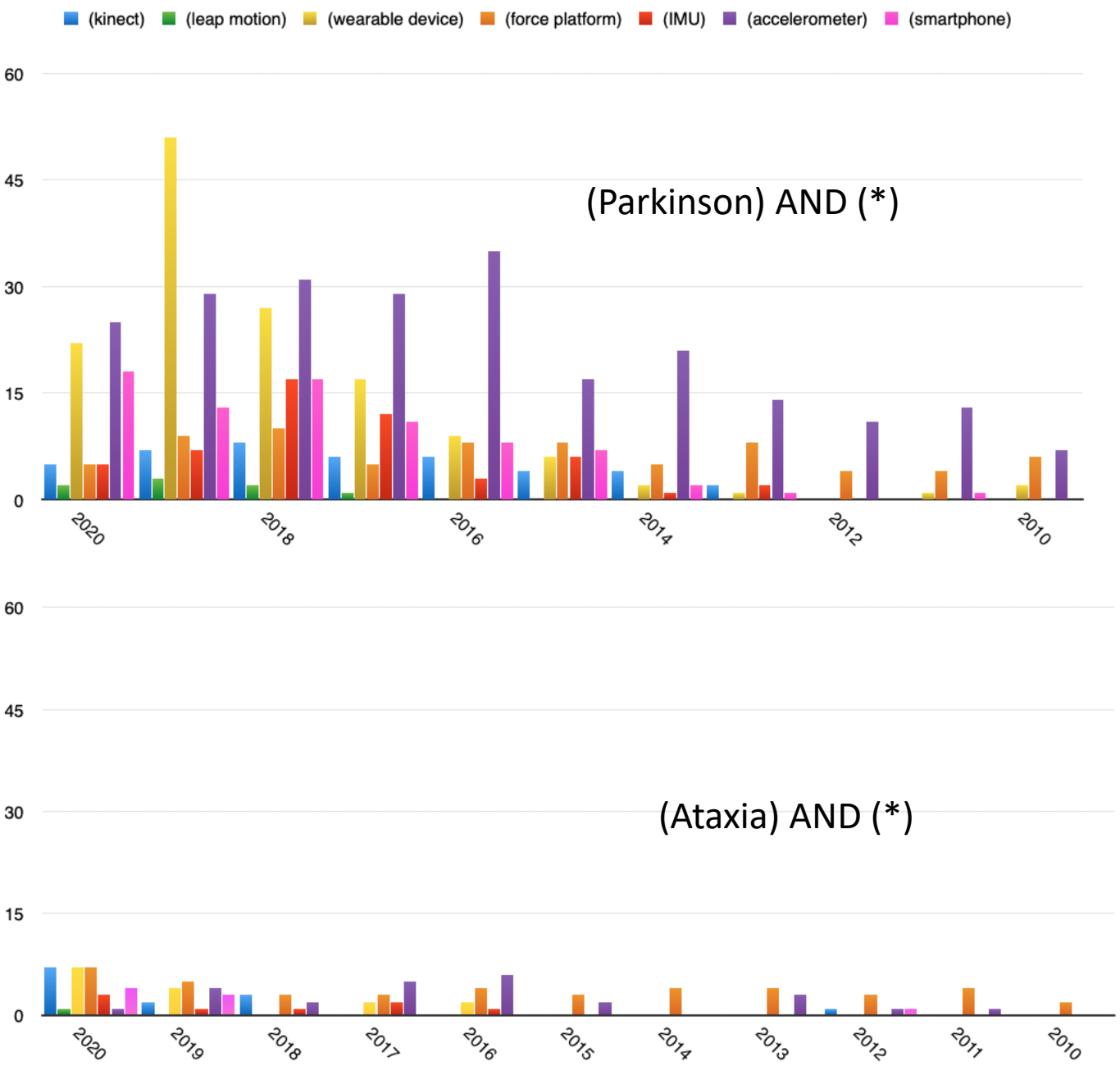
3) Store collected data

4) Data mining and analysis

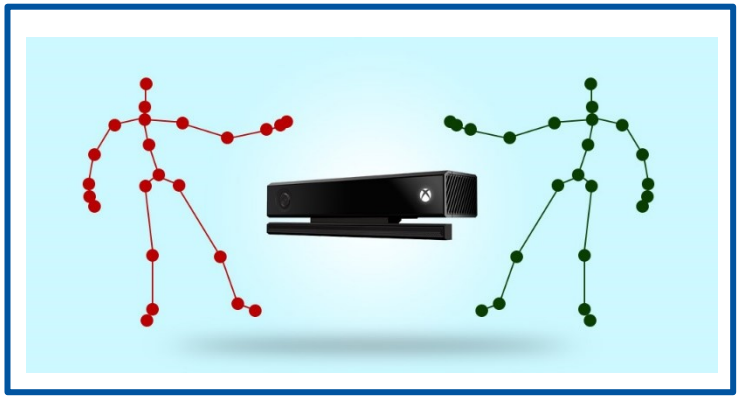
Database CNR
NO-SQL

Q5: Which devices would you choose developing a quantitative assessment system?

- Accelerometer
- Leap motion controller
- Force platform (posturography)
- Kinect
- Smartphone
- Wearable device
- IMU
- Other


(kinect)
(leap motion)
(wearable device)
(force platform)
(IMU)
(accelerometer)
(smartphone)
60
45
30
15
0
2020
2018
2016
2014
2012
2010
(Parkinson) AND (*)
60
45
30
15
0
2020
2019
2018
2017
2016
2015
2014
2013
2012
2011
2010
(Ataxia) AND (*)

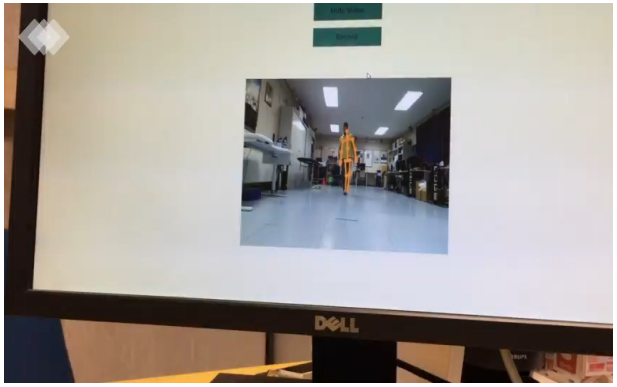
SARA@home: Optical sensors



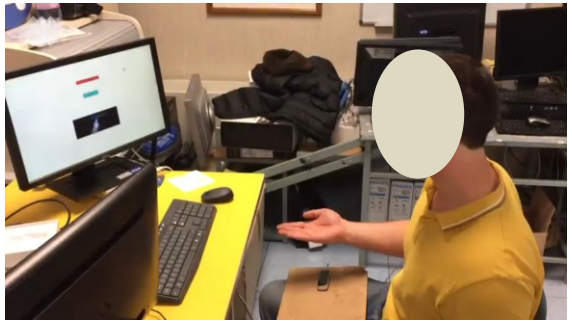
- **we wanted to record the movement as natural as possible**

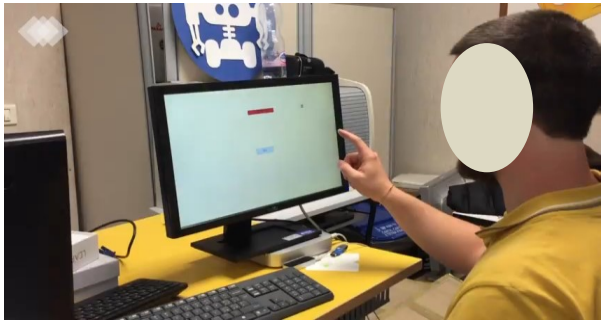
Our aim was to put in relationship the Kinect-Leap Motion data sequences and clinical SARA scale assessment

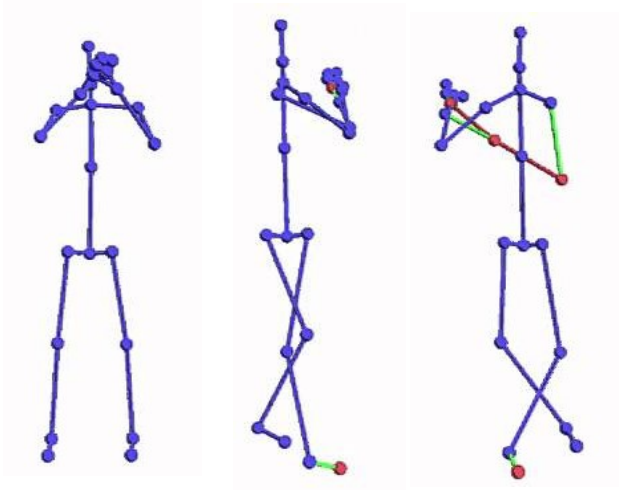
SARA@home: acquisition



Patients are **compliant and motivated** to complete tasks by the reproduction of his skeletal joint structure on the screen.







SARA@home: instructions

Video-guided:

A friendly actor placed into a home context strikes patient's attention without speaking (as a "teletubbies") and guides the tasks execution

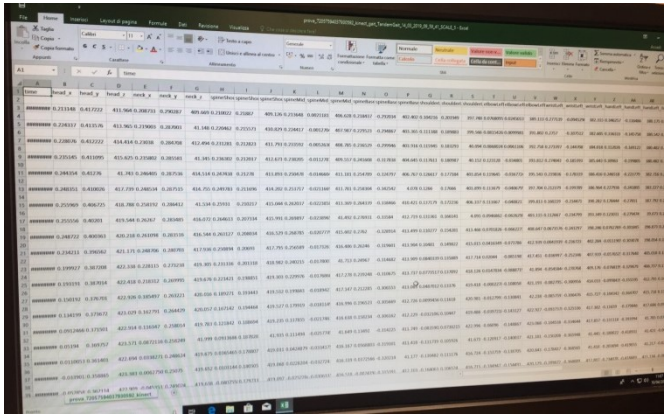
- To improve collaboration
- To allow assessment in absence of operators

SARA@home: analysis

Comparison and correlation

- Clinical data
- «VICON gait analysis» - gold standard

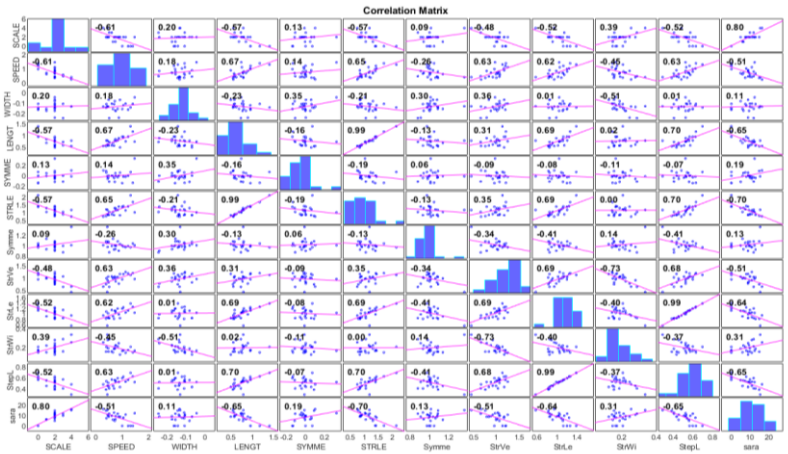
Data transformation



Computer Methods and Programs in Biomedicine
Volume 196, November 2020, 105705

Validation of low-cost system for gait assessment in children with ataxia

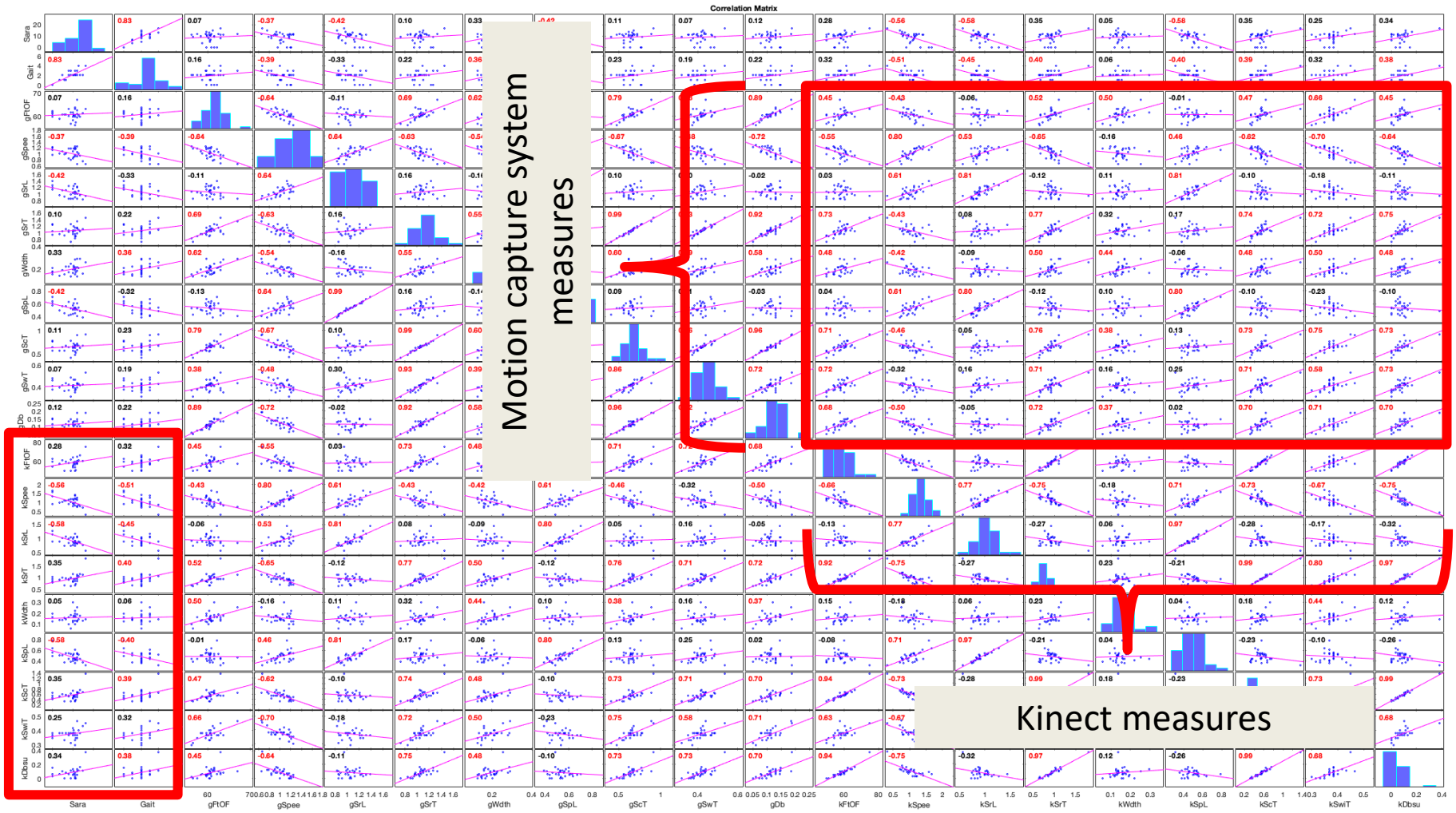
S. Summa [a], G. Tartarisco [b], M. Favetta [a], A. Buzachis [c], A. Romano [a], G.M. Bernava [b], A. Sancesario [a], G. Vasco [a], G. Pioggia [b], M. Petrarca [a], E. Castelli [a], E. Bertini [d], T. Schirinzi [a, e]



Computer Methods and Programs in Biomedicine
Volume 196, November 2020, 105705

SARA@home: analysis

Validation of low-cost system for gait assessment in children with ataxia



ELSEVIER

Computer Methods and Programs in Biomedicine

Volume 196, November 2020, 105705

SARA@home: analysis

Validation of low-cost system for gait assessment in children with ataxia

Table 2: Relationship between measures of each parameter acquired with the Kinect system with respect to those acquired with the motion capture system, reported in the first two columns. Bland-Altman test and correlation analysis between the parameters measured with the two systems.

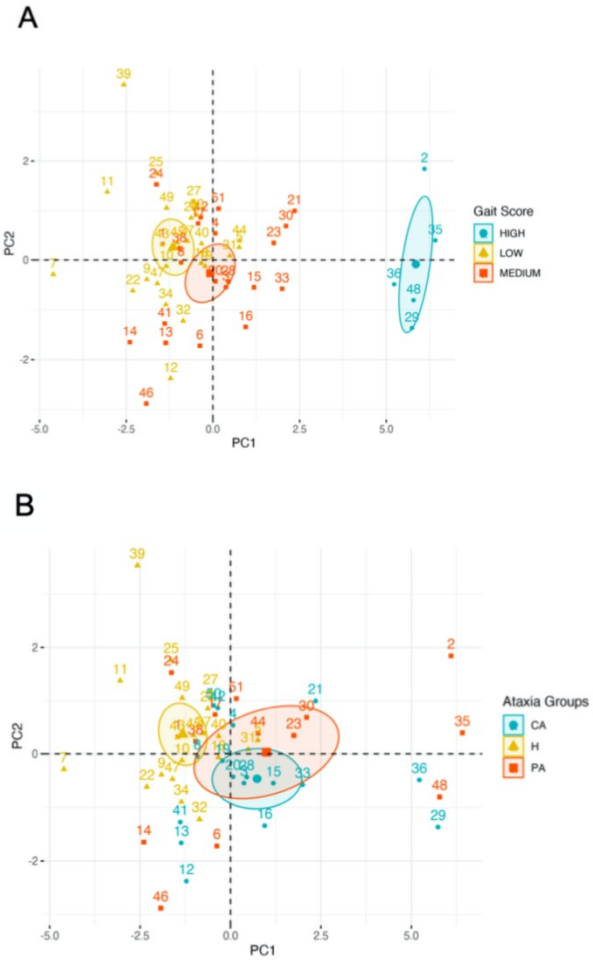
	r	p-value	Fixed bias	Proportional bias	LoA	RPC	Kinect Mean [CI]	Vicon Mean [CI]
Cadence [steps/min]	0.82	<0.0001	20*	0.31	[-12 51]	32	114.81 [82.81 146.81]	134.61 [83.02 86.21]
Speed [m/s]	0.80	<0.0001	0.04	0.31	[-0.3 0.37]	0.34	1.01 [0.68 1.34]	1.05 [0.51 1.59]
Stride Length [m]	0.81	<0.0001	-0.13*	-0.05	[-0.33 0.08]	0.20	1.06 [0.76 1.36]	0.93 [0.59 1.28]
Base Width [m]	0.44	0.01	-0.04*	-0.55*	[-0.13 0.06]	0.09	0.19 [0.11 0.28]	0.16 [0.07 0.25]
Step Length [m]	0.80	<0.0001	-0.05*	-0.03	[-0.16 0.06]	0.11	0.53 [0.38 0.68]	0.48 [0.30 0.67]
Stride Time [s]	0.77	<0.0001	-0.14*	0.11	[-0.4 0.13]	0.27	1.07 [0.78 1.35]	0.93 [0.51 1.35]
Stance Phase [%]	0.45	0.01	-2.3*	0.09	[-11 6.7]	9.1	60.7 [56.51 64.91]	58.4 [48.21 68.53]
Swing Phase [%]	0.46	0.01	-0.04*	0.10	[-6.7 11]	9.1	39.29 [35.07 43.52]	41.63 [31.47 51.79]
Double Support [%]	0.58	<0.0001	-0.03*	0.56	[-11 5.3]	7.9	10.63 [7.10 14.16]	7.99 [-1.42 17.41]

LoA = Limits of Agreement; RPC = reproducibility coefficient (1.96 x standard deviation); CI = Confidence Interval; r = Pearson coefficient; * p-value<0.05 given by the respective statistical test.

Table 3: Comparison of classification accuracy & Cohen's Kappa value [mean (IQR)%] with 10-fold cross validation.

Classifier	Low/Medium/High	PA/CA/H
MLP	67.5 (19) & 57 (12.1)	55.1 (5.5) & 30.4 (10.2)
NB	78.2 (20) & 61.5 (37)	51.1 (23.6) & 24.3 (34.6)
k-NN	68.5 (2.7) & 43.7 (5.8)	45.1 (10) & 14.2 (10.4)
RF	83.2 (0.9) & 72.8 (1.8)	58.9 (12.12) & 36.5 (15.8)
SVM	**90.4 (19) & 82.8 (34)**	**68.6 (3.4) & 49.7 (8.95)**

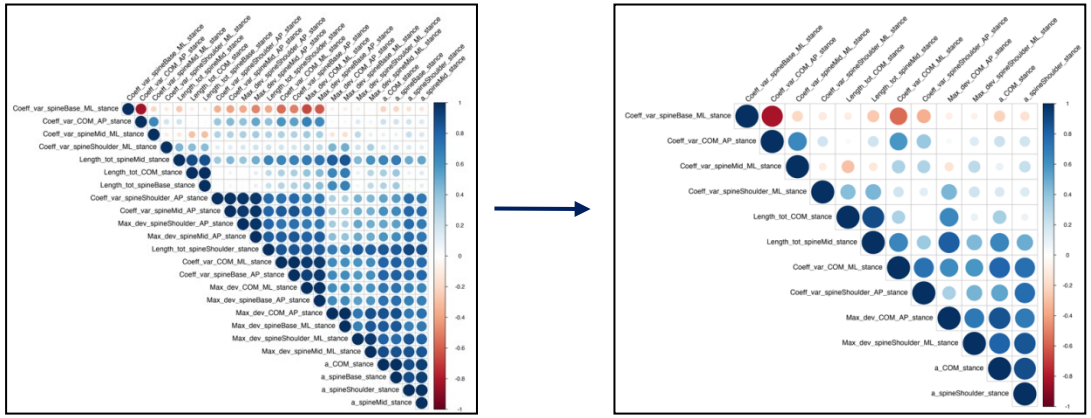
MLP = Multilayer Perceptron, NB = Naïve Bayes, k-NN = k-Nearest Neighbors, RF = random forest decision tree and SVM = support vector machine.



SARA@home: analysis

Signal processing → Features extraction → Features selection → Correlation analysis → Classifier analysis

Stance



	3 Classes: low - medium - high	70% training	5-fold
Fast hand	Features selection	55.6%	53.3%
	All Features	66.7%	46.7%

	3 Classes: low - medium - high	70% training	5-fold
Sitting	Features selection	86.7%	85.8%
	All Features	86.7%	85.8%

SARA@home: Machine learning results

We have selected those features - from all the items - that best correlates with SARA score

N Features	Features	Accuracy 5-fold
3	Stride_time | Stride_length | Step_width |	81.3%
7	PATA_freq | Stride_time | Stride_length | Pitch | mfcc1 | mfcc8 | mfcc13 |	89%
5	Stride_time | mfcc6 | mfcc10 | Stride_length | Spectral entropy |	88.9%
11	PATA_freq | stride_time | Spectral_entropy, mfcc10 | mfcc6 | stride_length | mfcc13 | mfcc7 | Zero-Crossings | mfcc1 | mfcc6 |	86.7%
11	SmoothY_r | PATA_freq | MeanAccX_r | VarAccZ_r | mfcc8 | Stride_length | MeanAccY_l | Stride_time | MeanAccZ_l | VarAccZ_l | mfcc1 |	88.57%
11	VarAccY_r | SmoothY_r | Zt_l | MeanAccZ_l | PATA_freq | mfcc7 | SmoothX_r | VarAccZ_r | mfcc8 | mfcc9 | Step_width |	92%

Journal of Neurology (2019) 266:1611–1622
https://doi.org/10.1007/s00415-019-09299-9

ORIGINAL COMMUNICATION

15-White Dots APP-Coo-Test: a reliable touch-screen application for assessing upper limb movement impairment in patients with cerebellar ataxias

Giuseppe Arcuria[1] · Christian Marcotulli[1] · Claudio Galasso[1] · Francesco Pierelli[1] · Carlo Casali[1]

Received: 26 January 2019 / Revised: 23 March 2019 / Accepted: 27 March 2019 / Published online: 6 April 2019

Journal of Neurology (2020) 267:625–639
https://doi.org/10.1007/s00415-019-09570-z

ORIGINAL COMMUNICATION

Developing a smartphone application, triaxial accelerometer-based, to quantify static and dynamic balance deficits in patients with cerebellar ataxias

Giuseppe Arcuria[1] · Christian Marcotulli[1] · Raffaele Amuso[2] · Giuliano Dattilo[3] · Claudio Galasso[1] · Francesco Pierelli[1,4] · Carlo Casali[1]

Received: 30 May 2019 / Revised: 3 October 2019 / Accepted: 9 October 2019 / Published online: 11 November 2019

- **APP-Coo-Test** is able to carry out quantitative and objective measurements of the rapid and coordinated upper limb movements and is also able to assess static and dynamic balance in patients (87) with cerebellar ataxias
- **15-White Dots APP** measurements have highly correlated with the scores obtained with the SARA, with the Composite Cerebellar Functional Severity (CCFS) and with the Nine Hole Pegboard test (9HPT) and the Click Test
- strong correlation between the **APP-Coo-Balance** measurements and the score obtained with the Berg Balance Scale, SARA, and a force platform (specific for posturography).
- the APP is an easy, reliable, and valid evaluating system to quantify the trunk sway in a static position and during the gait and to assess the severity of the upper limb ataxia

RESEARCH

A Comprehensive Scheme for the Objective Upper Body Assessments of Subjects with Cerebellar Ataxia

Ha Tran[1*], Khoa D Nguyen[1], Pubudu N Pathirana[1], Malcolm K Horne[2], Laura Power[3] and David J Szmulewicz[2,3,4]

Conferences > 2020 42nd Annual Internationa...

Multimodal Data Acquisition for the Assessment of Cerebellar Ataxia via Ballistic Tracking

Publisher: IEEE

Cite This PDF

Ha Tran; Khoa D. Nguyen; Pubudu N. Pathirana; Malcolm Horne; Laura Power; David J. Szmulewicz All Authors

- These studies assess upper-limb ataxia tests in patients (41) and controls (14) using motion measures obtained from a Kinect camera and a wearable motion-captured device (an IMU).
- The combination of multimodal features improved the ability to distinguish (using PCA and machine learning models) between patients and controls and to measure the severity of upper limb ataxia.
- model accuracy is 96% and correlation with clinical scores is 80%

The Cerebellum
https://doi.org/10.1007/s12311-020-01210-x

ORIGINAL ARTICLE

The Use of New Mobile and Gaming Technologies for the Assessment and Rehabilitation of People with Ataxia: a Systematic Review and Meta-analysis

Eleonora Lacorte[1] · Guido Bellomo[1] · Sara Nuovo[2] · Massimo Corbo[3] · Nicola Vanacore[1] · Paola Piscopo[4]

Accepted: 1 November 2020

«In this review, the authors assessed currently available evidence on the use of new mobile and gaming technologies in the assessment and rehabilitation of people with chronic ataxias.
…
We found only 2 diagnostic studies investigating the use of these technologies for the assessment of specific motor functions in people with chronic ataxias. Though having an overall low-quality score, they both **reported these tools to be useful and reliable**. The low quality of these studies was mainly due to their being designed as case-control diagnostic studies and the enrollment of subjects with different diagnoses, disease duration, and degree of severity. **The rarity of the disease, however, makes it virtually impossible to design conventional diagnostic studies.**
…
However, **adopting a multicenter approach and involving organizations of ataxic patients could allow enrolling a larger number of participants**, increasing the size of subgroups with homogeneous phenotypes.»

Key Points /Conclusions

- Need to define and to standardize measures (= > digital/bio-markers)
- The choice of which technology to use is related to the contest
- Acceptance of technology is fundamental for a home-based tool
- So far studies are isolated experience with technologies used only by its original developer while multicentric studies are needed (rare disease)

We are in line with the current methodologies and results obtained looking at baseline

Moreover… longitudinal observation (2 follow-ups) of:

- 21 patients with progressive ataxia
- 17 patients with non progressive/congenital ataxia
- 21 healthy controls

We are looking at the sensitivity to change of the SARA@home assessment

Enrico Bertini

Gessica Vasco

Ginevra Zanni

Enrico Castelli

MARlab

THANK YOU

Co-financed by the Connecting Europe Facility of the European Union

European Reference Network for rare or low prevalence complex diseases
Network Neurological Diseases (ERN-RND)

European Reference Network for rare or low prevalence complex diseases
Network Neuromuscular Diseases (ERN EURO-NMD)

European Reference Network | Neurological Diseases (ERN-RND)

This webinar has been supported by ERN-RND , which is partly co-funded by the European Union within the framework of the Third Health Programme "ERN-2016 - Framework Partnership Agreement 2017-2021."

DG ‚Ataxia and HSP'
24. November 2020

Joint webinar series

THANK YOU

Next Webinar: 1. December 2020

‚Functional movement disorders: a diagnostic guide'

by Christos Ganos,

Charité University Medicine, Berlin, Germany