Doença de Huntington

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Huntington's Disease: Present Treatments and Future Therapeutic Modalities

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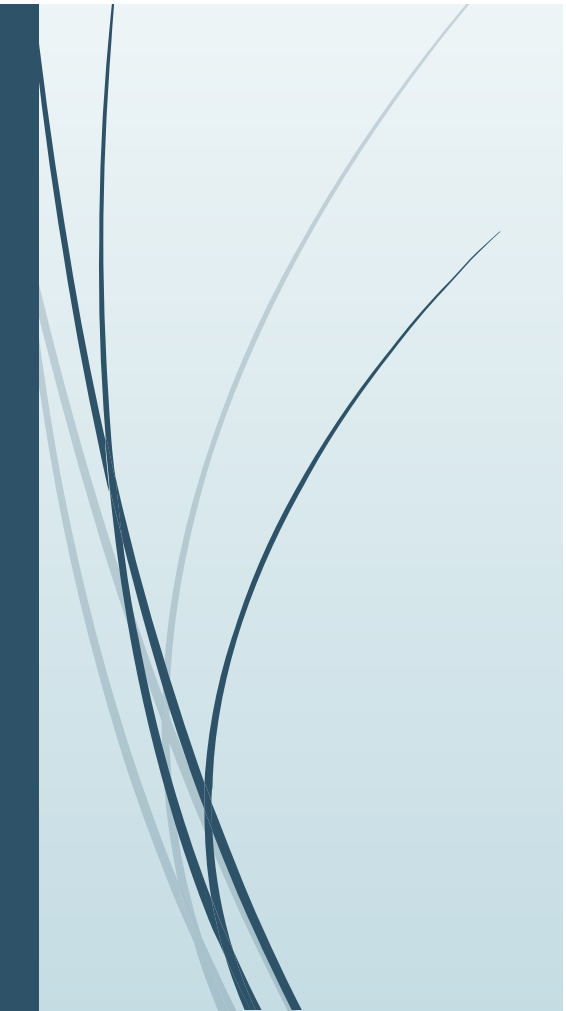

Abstract

Huntington's disease (HD) is a devastating neuropsychiatric disorder for which therapeutic interventions have been rather fruitless to date, except in a slight symptomatic relief. Even the discovery of the gene related to HD in 1993 has not effectively advanced treatments. This article is essentially a review of available double-blind, placebo-controlled trials of therapy for this condition which also includes relevant open label trials. Unfortunately, HD research has tended to concentrate on the motor aspects of the disorder, whereas the major problems are behavioural (e.g. dementia, depression, psychosis), and the chorea is often least relevant in terms of management. We conclude that there is definitely poor evidence in management of HD. The analysis of the 24 best studies fails to result in a treatment recommendation of clinical relevance. Based on data of open-label studies, or even case reports, we recommend riluzole, olanzapine and amantadine for the treatment of the movement disorders associated with HD, selective serotonin reuptake inhibitors and mirtazapine for the treatment of depression, and atypical antipsychotic drugs for HD psychosis and behavioural problems. Moreover, adjuvant psychotherapy, physiotherapy and speech therapy should be applied to supply the optimal management. Finally, some cellular mechanisms are discussed in this paper because they are essential for future neuroprotective modalities, such as minocycline, unsaturated fatty acids or riluzole.

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[Telemonitoring of Swallowing Function: Technologies in Speech Therapy Practice]

[Article in Italian]

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Abstract

The process of medical-healthcare technological revolution represents an advantage for the patient and for the care provider, in terms of costs and distances reduction. The telehomecare approach could be useful for monitoring the swallowing disorder in neurodegenerative diseases, preventing complications. In this study the applicability of telemedicine techniques for the monitoring of swallowing function, in patients affected by Huntington's disease (HD), was evaluated through the acquisition and analysis of the sound of swallowing. Two patients with HD were outpatient screened for dysphagia through the Bedside Swallowing Assessment Scale (BSAS) sensitized with pulse oximetry and cervical auscultation. Subsequently, the swallowing functionality was telemonitored for three months with Skype. The swallowing sounds were acquired with a detection microphone attached to the lateral edge of the trachea during fluid intake. The sounds were instantly processed and graphically represented through the Praat software. The analysis of the acoustic signal acquired remotely has made it possible to identify the situations that required immediate speech therapy intervention, suggesting to the patients further modifications of food consistencies, and saving frequent moving to the hospital even in the absence of critical situations. Remote assistance applied to speech therapy could represent a benefit for patients and their carers and a more efficient use of medical and health resources.

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Tremor and Other Hyperkinetic Movements

Reviews

Speech–Language Pathology Evaluation and Management of Hyperkinetic Disorders Affecting Speech and Swallowing Function

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Abstract

Background: Hyperkinetic dysarthria is characterized by abnormal involuntary movements affecting respiratory, phonatory, and articulatory structures impacting speech and deglutition. Speech–language pathologists (SLPs) play an important role in the evaluation and management of dysarthria and dysphagia. This review describes the standard clinical evaluation and treatment approaches by SLPs for addressing impaired speech and deglutition in specific hyperkinetic dysarthria populations.

Methods: A literature review was conducted using the data sources of PubMed, Cochrane Library, and Google Scholar. Search terms included 1) hyperkinetic dysarthria, essential voice tremor, voice tremor, vocal tremor, spasmodic dysphonia, spastic dysphonia, oromandibular dystonia, Meige syndrome, orofacial, cervical dystonia, dystonia, dyskinesia, chorea, Huntington's Disease, myoclonus; and evaluation/treatment terms: 2) Speech–Language Pathology, Speech Pathology, Evaluation, Assessment, Dysphagia, Swallowing, Treatment, Management, and diagnosis.

Results: The standard SLP clinical speech and swallowing evaluation of chorea/Huntington's disease, myoclonus, focal and segmental dystonia, and essential vocal tremor typically includes 1) case history; 2) examination of the tone, symmetry, and sensorimotor function of the speech structures during non-speech, speech and swallowing relevant activities (i.e., cranial nerve assessment); 3) evaluation of speech characteristics; and 4) patient self-report of the impact of their disorder on activities of daily living. SLP management of individuals with hyperkinetic dysarthria includes behavioral and compensatory strategies for addressing compromised speech and intelligibility. Swallowing disorders are managed based on individual symptoms and the underlying pathophysiology determined during evaluation.

Discussion: SLPs play an important role in contributing to the differential diagnosis and management of impaired speech and deglutition associated with hyperkinetic disorders.

Keywords: Hyperkinetic dysarthria, dystonia, dyskinesia, chorea, myoclonus, essential vocal tremor

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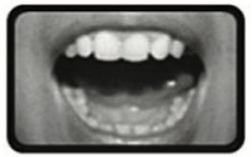
CASE HISTORY

- Determine the degree of impact of symptoms on the patient's daily life activities including occupational, social, family, etc.
- Determine duration of symptoms and changes in symptoms from onset to current time
- Elicit patient description of their symptoms for speech and non-speech activities
- Determine the impact of symptoms on the ability to be understood (i.e. intelligibility)
- Inquire about factors that reduce or worsen involuntary movements during voluntary activities
- Acquire patient's sense of effort associated with speaking and deglutition
- Determine level of awareness of patient regarding abnormal movements triggered by volitional activities (e.g. speaking, chewing food, posturing of orofacial structures, etc.)
- Determine methods the patient has already attempted to manage symptoms and their current goals

QUALITY OF LIFE SELF-REPORT

- Administration of currently available instrument related to assessing the impact of the individual's dysarthria or voice symptoms on their ability to participate in daily life activities



MOTOR SPEECH ASSESSMENT: ORAL MECHANISM ASSESSMENT

- Observe the tone and symmetry of the orofacial structures (e.g. face, jaw, tongue, velum)
- Test the strength, range of motion, and speed of movements of the orofacial structures
- Test reflex integrity (e.g. gag response, jaw jerk, snout reflex, sucking reflex)
- Test coordination and integrity of speech structures and subsystems needed to perform a cough, puffing out the cheeks with air, smacking and licking the lips, blowing, clicking the tongue, and biting the lower lip

MOTOR SPEECH ASSESSMENT: SPEECH ASSESSMENT

- Test respiratory-phonatory coordination, voice quality, and resonance during maximum voicing duration (e.g. sustained "ah")
- Test diadochokinetic rate and phoneme integrity during single, double, and multi-syllable repetition
- Evaluate functional speech during standard passage and sentence reading and conversational speech
- Assess intelligibility and communication efficiency at the word and sentence level

Figure 1. **Components of a speech motor evaluation**. This figure describes the typical activities associated with the case history, quality of life self-report, and motor speech assessment (oral mechanism and speech assessment portions).

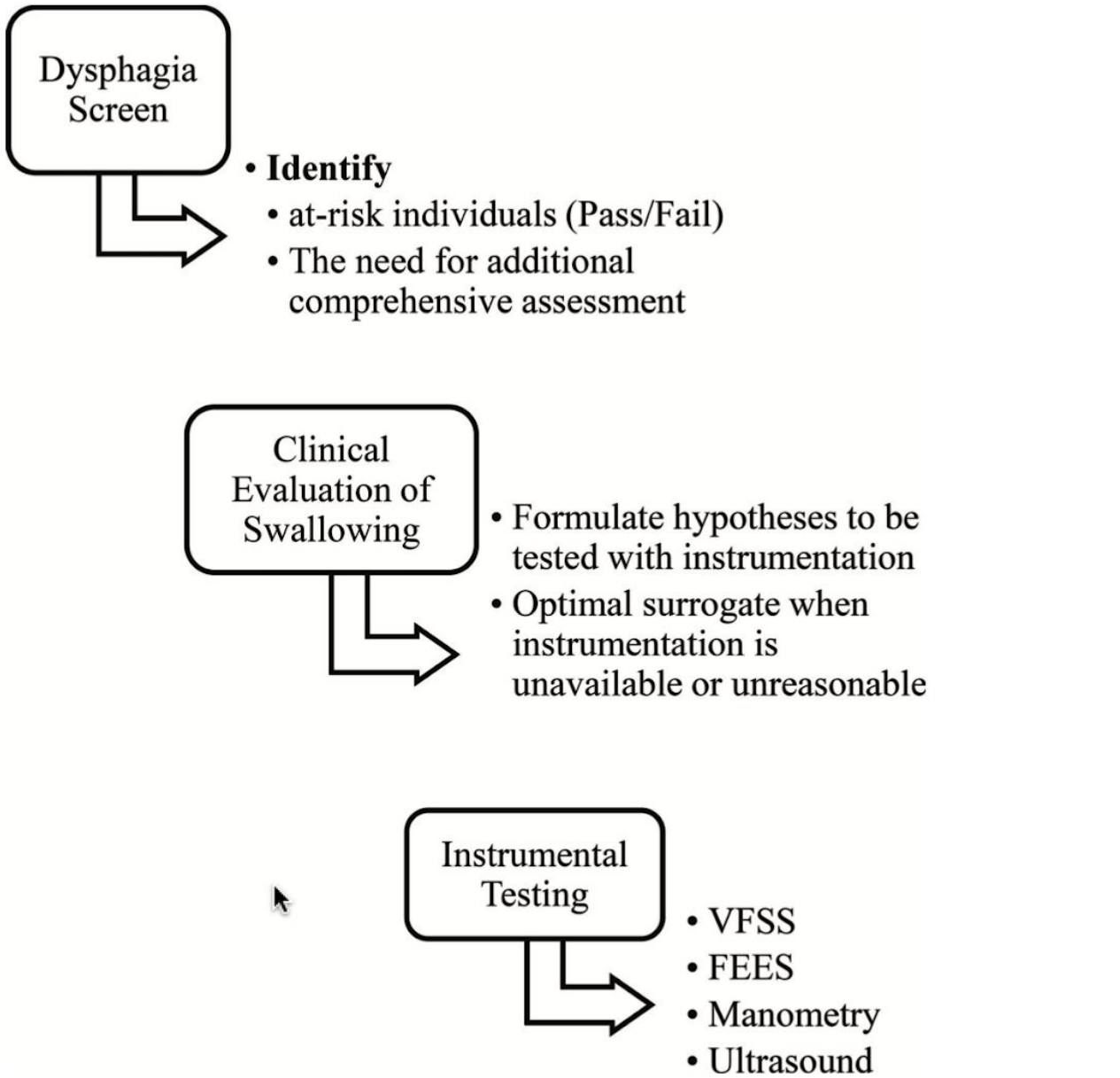


Figure 4. **Process for identifying and evaluating dysphagia**. This figure illustrates the recommended clinical practice pattern for speech-language pathologist identification and evaluation of individuals with dysphagia.

Table 4. **Speech–Language Pathologist Treatment Approaches to Managing Impaired Respiratory, Voice, and Articulatory Functions in Those with Hyperkinetic Dysarthria**

Hyperkinetic Dysarthria Characteristic	Sign/Symptom	Treatment Options
Impaired respiratory drive, or coordination for speech production	Reduced or inconsistent loudness	Expiratory muscle strength training
	Dramatic reduction in loudness during a single breath group during speaking	Lee Silverman Speech Treatment (LSVT)
	Inhalation appears inadequate, prolonged, or speaking initiation occurs at unusual locations within the respiratory cycle, or utterance	Maximum inhalation/exhalation tasks, or sustained phonation tasks to improve respiratory/phonatory coordination and steadiness
	Few words or syllables produced per breath group, runs out of air before taking a breath	Body positioning to optimize breathing and respiratory efficiency during speaking
	Paradoxical movements of the rib cage and abdomen during breathing or speaking	Accent Method of Voice Therapy
	Abnormal posture or movements associated with volitional respiratory-phonatory coordination during speaking	Rehearse taking deeper inhalations prior to speaking and implementing increased respiratory effort during speaking
	Reduced maximum phonation time (may also indicate impaired voice function)	Rehearse optimal breath groups during phrasing of spoken utterances
Impaired voice function	Poor integrity, loudness, and rate of laryngeal diadochokinesis (e.g., ee-ee-ee-ee)	Laryngeal relaxation techniques such as easy voice onset, yawn-sign, chanting, chewing method
		Laryngeal Manipulation
		Accent Method of Voice Therapy
		Confidential Voice Technique/Flow Phonation
	Hyperadduction of the vocal folds	Biofeedback during voicing/speech tasks
Impaired speech function	Impaired articulation	Articulation therapy
		Modify speaking rate (typically encourage slower)
	Abnormal speech pattern or rate	Speech rhythm techniques
	Abnormal resonance (e.g., hypernasality)	Delayed auditory feedback
		Direct magnitude production
		Augmentative and alternative communication intervention
		Referral for prosthetic device

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HDQLIFE: The development of two new computer adaptive tests for use in Huntington disease, Speech Difficulties and Swallowing Difficulties

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Abstract

Purpose—Huntington disease (HD) is an autosomal dominant neurodegenerative disease which results in several progressive symptoms, including bulbar dysfunction (i.e., speech and swallowing difficulties). Although difficulties in speech and swallowing in HD have a negative impact on health-related quality of life, no patient-reported outcome measure exists to capture these difficulties that are specific to HD. Thus, we developed a new patient reported outcome measure for use in the Huntington Disease Health-Related Quality of Life (HDQLIFE) Measurement System that focused on the impact that difficulties with speech and swallowing have on HRQOL in HD.

Methods—Five hundred seven individuals with prodromal and/or manifest HD completed 47 newly developed items examining speech and swallowing difficulties. Unidimensional item pools were identified using exploratory and confirmatory factor analyses (EFA and CFA, respectively). Item response theory (IRT) was used to calibrate the final measures.

Results—EFA and CFA identified two separate unidimensional sets of items: Speech Difficulties (27 items) and Swallowing Difficulties (16 items). Items were calibrated separately for these two measures and resulted in item banks that can be administered as computer adaptive tests (CATs) and/or 6-item, static short forms. Reliability of both of these measures was supported through high correlations between the simulated CAT scores and the full item bank.

Conclusions—CATs and 6-item calibrated short forms were developed for HDQLIFE Speech Difficulties and HDQLIFE Swallowing Difficulties. These measures both demonstrate excellent psychometric properties, and may have clinical utility in other populations where speech and swallowing difficulties are prevalent.

Keywords

Health-related quality of life; Neuro-QoL; PROMIS; HDQLIFE; Huntington disease; Speech difficulties; Swallowing difficulties; Patient reported outcome (PRO)

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HDQLIFE: development and assessment of health-related quality of life in Huntington disease (HD)

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Abstract

Purpose Huntington disease (HD) is a chronic, debilitating genetic disease that affects physical, emotional, cognitive, and social health. Existing patient-reported outcomes (PROs) of health-related quality of life (HRQOL) used in HD are neither comprehensive, nor do they adequately account for clinically meaningful changes in function. While new PROs examining HRQOL (i.e., Neuro-QoL—Quality of Life in Neurological Disorders and PROMIS—Patient-Reported Outcomes Measurement Information System) offer solutions to many of these shortcomings, they do not include HD-specific content, nor have they been validated in HD. HDQLIFE addresses this by validating 12 PROMIS/Neuro-QoL domains in individuals with HD and by using established PROMIS methodology to develop new, HD-specific content.

Methods New item pools were developed using cognitive debriefing with individuals with HD, and expert, literacy, and translatability reviews. Existing item banks and new item pools were field tested in 536 individuals with prodromal, early-, or late-stage HD.

Results Moderate to strong relationships between Neuro-QoL/PROMIS measures and generic self-report measures of HRQOL, and moderate relationships between Neuro-QoL/PROMIS and clinician-rated measures of similar constructs supported the validity of Neuro-QoL/PROMIS in individuals with HD. Exploratory and confirmatory factor analysis, item response theory, and differential item functioning analyses were utilized to develop new item banks for Chorea, Speech Difficulties, Swallowing Difficulties, and Concern with Death and Dying, with corresponding six-item short forms. A four-item short form was developed for Meaning and Purpose.

Conclusions HDQLIFE encompasses both validated Neuro-QoL/PROMIS measures, as well as five new scales in order to provide a comprehensive assessment of HRQOL in HD.

Keywords Neuro-QoL · PROMIS · Health-related quality of life · HDQLIFE · Huntington disease · Patient-reported outcome (PRO)

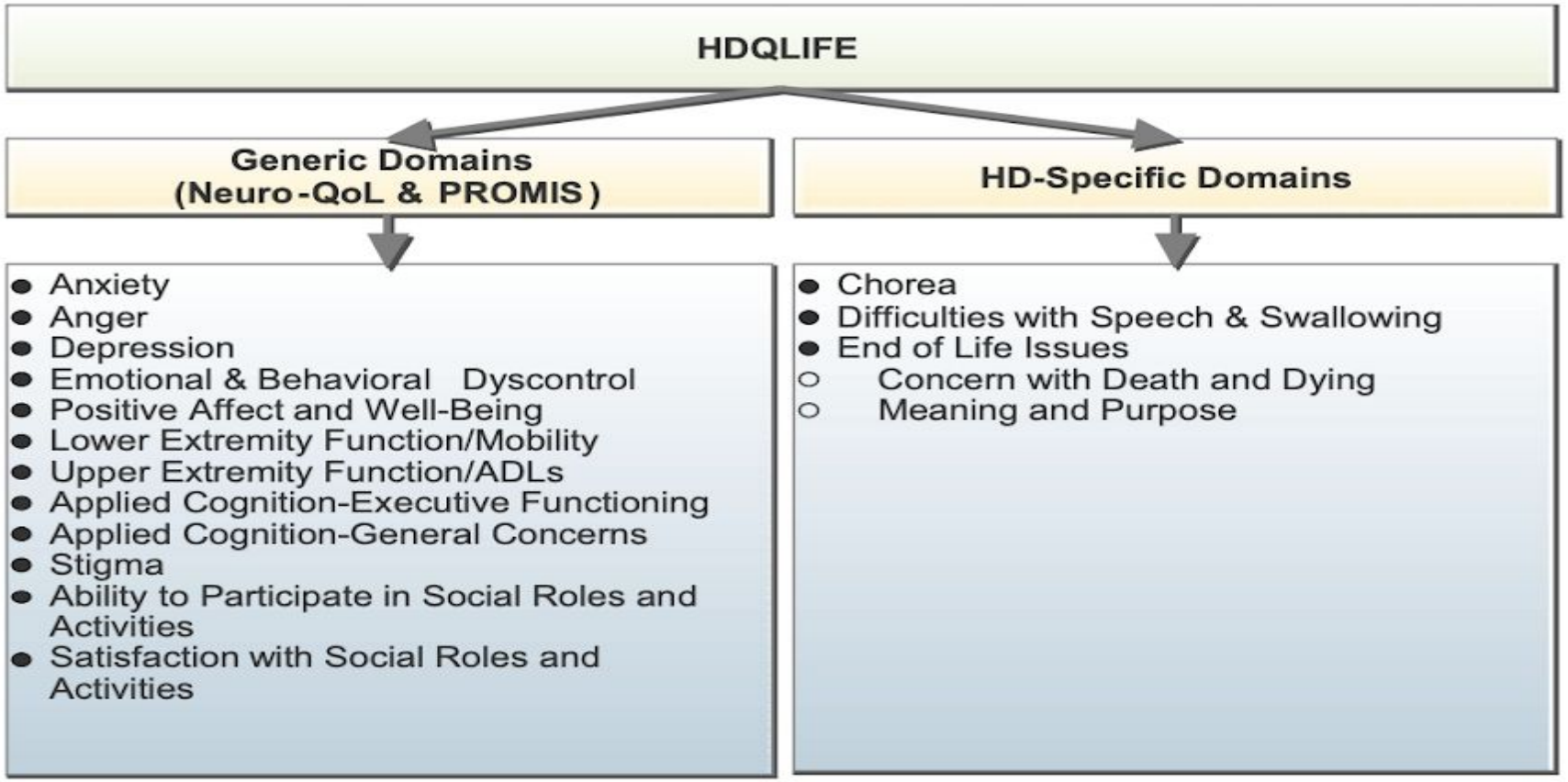

Fig. 1 Components of the HDQLIFE measurement system