HEALTH AND SOCIAL CARE IN THE COMMUNITY (2019) – Pre-print

Raffaela DG Sartori, Marco Marelli, Maria Grazia D'Angelo, Antonella Delle Fave

Autonomy level and quality of everyday experience of people with Hereditary Spastic Paraplegia

Abstract

In the health domain, well-being is primarily assessed as autonomy and mental distress, while the quality of daily experience is rarely investigated. In the present study, the relationship between autonomy levels and daily experience was explored.

Thirty-five Italian adults with Hereditary Spastic Paraplegia provided for one week real-time descriptions of daily activities and associated experiences through the Experience Sampling Method procedure. Participants were grouped based on autonomy levels assessed through Barthel Index. The relationships between activity typologies, the experiential dimensions of perceived challenges and skills, and autonomy level were analyzed.

Participants' predominant activities were personal care, associated with global disengagement, and leisure, associated with high control and desirability, but low perceived relevance. During social interactions participants reported engagement and emotional well-being, and during productive activities high activation but negative affect. Multi-level analysis highlighted that this association between activity type and experiential patterns recurred across autonomy levels. In addition, perceived challenges in the activity were lower that perceived personal skills across activities and autonomy levels. Findings suggest that persons with motor disabilities, regardless of their autonomy level, would benefit from more challenging opportunities for action in daily life, in order to attain well-being through active skill mobilization.

Keywords

Neuromuscular diseases, Hereditary Spastic Paraplegia, Experience Sampling Method, Quality of experience

Bullet points

What is known about this topic

- The daily experience of persons with disabilities is rarely analyzed through real-time procedures
- Physical health conditions are only partially related to individuals' daily quality of experience
- The interplay between perceived environmental challenges and personal skills is a key determinant of the quality of experience

What this paper adds

- Participants' autonomy level had a modest impact on the experience associated with daily activities
- The level of complexity and structure of daily activities were good predictors of participants' quality of experience
- The daily life of persons with disabilities primarily includes unstructured, passive and low-challenge tasks, thus entailing risks for their psychophysical health

1. INTRODUCTION

1.1. Theoretical Framework

The Italian National Registry for Rare Diseases (Taruscio, Kodra, Ferrari et al., 2014) includes people diagnosed with Spastic Paraparesis (Hereditary Spastic Paraplegias - HSPs), a neuromuscular disease affecting 1.3-12.1/100.000 citizens. In Italy 19,9 cases on 100.000 are estimated (17,5/100.000 for autosomal dominant forms, Racis, Tessa, Di Fabio et al., 2014); disease prevalence however varies across geographic areas and according to diagnosis and classification criteria (Ruano, Melo, Silva et al., 2014).

HSPs represent a heterogeneous group of inherited neurodegenerative disorders, including over 70 genetic subtypes with Mendelian inheritance (autosomal dominant, autosomal recessive, X-linked) and non-Mendelian mitochondrial maternal transmission (Lo Giudice, Lombardi, Santorelli et al., 2014). The most common pathological feature of HSPs is a retrograde distal axonopathy of the longest descending motor fibers of the corticospinal tract and posterior columns (Blackstone, O'Kane & Reid, 2011). The main clinical manifestations are lower limb spasticity and weakness (Klebe, Stevanin & Depienne, 2015). Disease progression is usually slow, and it can even evolve into stabilization in some forms arising during childhood, but clinical variability can be observed in forms sharing the same mutation (Fink, 2003).

Currently, therapies are limited. Physical treatments and antispastic drugs help reduce muscle spasticity and bladder dysfunction (McDermott, White, Bushby et al., 2000). Regular physiotherapy cycles contribute to improving lower limbs' muscle strength and cardiovascular function (Fink, 2003).

Besides medical aspects, persons with HSPs face challenges in daily activity management, psychological adjustment, and fulfillment of social roles. These issues need to be addressed through an integrated approach, overcoming the biomedical and deficit-centered view. This approach is endorsed by the International Classification of Functioning, Disability and Health (ICF, WHO, 2001), that

conceptualizes disability as a variation of human functioning with biological, personal and social dimensions. Research on the daily management of neuromuscular diseases is increasingly focused on the investigation of psychological resources (Pagnini, 2013; Putnam, Greenen & Powers, 2003) such as adaptive coping strategies, that represent a key asset for attaining good quality of life and social integration (Simmons, Bremer, Robbins et al., 2000; Piccininni, Falsini, & Pizzi, 2004).

Challenges and resources perceived in disease conditions can be fruitfully explored through the study of daily subjective experience (Folkman & Moskowitz, 2000), a complex phenomenon emerging from the moment by moment interaction between environmental stimuli - such as daily activities and contexts - and cognitive, emotional and motivational dimensions (Hektner, Schmidt & Csikszentmihalyi, 2007). The relationship between daily activities and the associated experience can be operationalized in terms of balance or imbalance between the challenges perceived in the ongoing task and the personal skills perceived in facing these challenges (Fullagar, Knight & Sovern, 2013). A vast literature highlights that structured tasks in any domain (work, study, leisure, socializing) are associated with the perception of high challenges, that promotes skill mobilization and the onset of positive states such as flow or optimal experience, a rewarding state of engagement and absorption in the task at hand (Csikszentmihalyi, 1975/2000; Massimini & Delle Fave, 2000). To the contrary, unstructured or repetitive tasks like personal maintenance or watching TV are usually associated with the perception of low challenges, leading to negative experiences of disengagement (Kubey & Csikszentmihalyi, 1990; Delle Fave & Massimini, 2005a).

1.2. Aims and hypotheses

This study was aimed at investigating the quality of experience people with HSPs associate with daily activities, in order to verify the potential of these activities to promote positive experiences, including flow. To this purpose, persons with HSPs were administered the Experience Sampling

Method (ESM, Hektner et al., 2007), a real-time sampling procedure characterized by the repeated recording of daily activities and associated experiences. The preliminary assessment of autonomy level in Activities of Daily Living (ADL), an indicator of the person's potential to access environmental affordances, led to the identification of three groups of participants with low, moderate and high autonomy levels respectively.

The following hypotheses were formulated:

H1) regardless of autonomy level, participants' quality of experience would differ across daily activity types. Highly structured activities, such as productive tasks, would be associated with higher challenges and engaging experiences than simple and maintenance tasks, such as passive entertainment and personal care. Interpersonal interactions, representing opportunities for social integration, would be associated with globally positive experiences at the cognitive, emotional and motivational levels;

H2) more autonomous participants would report an overall better quality of experience than less autonomous ones across activity types;

H3) a specific interplay pattern between perceived challenges and skills would emerge based on activity type and autonomy level. More specifically, during productive activities and social interactions (a) all participants would report higher challenges and skills than in the other activities, and (b) highly autonomous participants would report higher values of both variables compared to the other two groups.

2. METHOD

2.1. Study design and contexts

Through a cross-sectional design, information was collected among people with HSPs about their daily activities and associated quality of experience in real time, during one standard week, considering participants' functional independence in activities of Daily Living (ADL).

Participants were recruited in collaboration with the Scientific Institute IRCCS "Eugenio Medea", that provides healthcare, rehabilitation and psychosocial services to people with neurological diseases; and the Italian Association of Persons with HSPs (Associazione Italiana Vivere la Paraparesi Spastica – A.I.Vi.P.S.). After project approval by the Ethics Committee of IRCCS Medea, potential participants were selected by the Institute medical staff during hospitalization for routine check-up or rehabilitation trainings, and by the A.I.Vi.P.S board by exploring the availability of members satisfying the inclusion criteria.

People who expressed interest in participating were contacted by the researchers and took part in the study between May 2009 and December 2012. Participants' autonomy level was assessed by their reference physicians through the Barthel Index (BI, Mahoney & Barthel, 1965).

2.2. Participants

To fulfill the study aims, participant selection was based on functional independence in ADL. Due to the rare occurrence and uneven geographic distribution of HSPs (Ruano et al., 2014), the sample size could not be defined according to statistical power considerations, leading to the forceful adoption of a non-probabilistic sampling. This approach is consistent with the guidelines included in "REporting of studies Conducted using the Observational Routinely collected health Data" (RECORD, Benchimol, Smeeth, Guttmann et al., 2015), which concern routinely collected health data, obtained for administrative and clinical purposes without specific a priori research goals

Participants were adults clinically diagnosed with Hereditary Spastic Paraplegia, who could write and complete the questionnaires by themselves. Exclusion criteria comprised moderate to severe mental retardation (QI<55, Wechsler, 1981) and diagnosed psychiatric disorders.

During a briefing session, participants received detailed information about the study structure and procedure; after signing an informed consent form, they received a booklet of Experience Sampling Method questionnaires and an electronic device programmed to send random signals for one week. They completed a sample form on site, so that related questions and doubts could be discussed. During a debriefing session, scheduled after the data collection week, participants returned the study materials and provided their comments. Participants were all volunteers; they were free to leave the study at any time, and their anonymity was guaranteed in all research phases.

2.3. Sources of data collection

The Barthel Index (BI, Mahoney & Barthel, 1965) measures a person's autonomy level in performing daily activities through 10 items, referring to personal hygiene, feeding, controlling bowels and bladder, and ability to move with or without aids. The sum of all item scores represents the functional independence index or BI (0-100), with scores in the 0-20 range corresponding to total dependence in ADL; scores 21-60 severe dependence; scores 61-90 moderate independence; and scores above 90 total independence (Shah, Vanclay & Cooper, 1989). Based on their BI score, participants were included in the low autonomy (LBI), moderate autonomy (MBI) or high autonomy (HBI) group.

Experience Sampling Method (ESM, Hektner et al., 2007) is designed to assess subjective experience in real time, during the unfolding of daily events. Participants receive randomized acoustic signals 6-8 times a day between 8 a.m. and 10 p.m. through an electronic device. At each signal reception, they are expected to complete a form containing open-ended questions, Likert-type 0–12 scales, and two questions recording the time of signal reception and the time of form completion. Open

ended questions investigate participants' activities, location and company at signal reception. Scaled questions measure the level of cognitive, affective and motivational dimensions of the experience, as well as perceived environmental challenges and personal skills. To the purpose of this study the description of the ongoing activity was selected for analysis, together with the following experiential variables: concentration and control (cognitive dimensions); feeling happy and active (emotional dimensions); stakes in the activity and activity desirability (motivational dimensions).

In line with previous studies involving participants diagnosed with psychiatric disorders (Bassi, Ferrario, Ba et al., 2012), participants with mild mental retardation (QI range 55-70) were provided with shorter ESM forms investigating the ongoing activity, the time of signal reception and form completion, and the ratings of fewer experiential variables, including those selected for the present study.

The validity and reliability of Experience Sampling Method (ESM) were widely investigated (Delle Fave et al., 2011; Hektner et al. (2007). The instrument was used across cultures, with clinical and non-clinical samples, in cross-sectional and longitudinal investigations (Bassi et al., 2012; Conner, Tennen, Fleeson, & Feldman Barrett, 2009; Mill, Kööts-Ausmees, Allik, & Realo, 2018; Trull & Ebner-Priemer, 2009).

Compared to single-administration procedures, ESM combines the ecological validity of naturalistic observation with the descriptive nature of diaries and the precision of scaled questionnaires, by allowing researchers to simultaneously gather information on individuals' behavior, environment and subjective experience, and to explore their interplay in real life settings (Hektner et al., 2007; Napa Scollon, Kim-Prieto, & Diener, 2003).

As concerns ESM response rate, a proportion of 30% of signals for which each participant completed a form is considered as adequate for inclusion in analyses (Hetkner et al., 2007). Across studies, response rate varied according to sample characteristics, ranging from 92% among US

managers (Csikszentmihalyi and Larson, 1987) to 68.8% among Italian teenagers (Bassi & Delle Fave, 2012). Response rates between 50% and 60% were obtained among persons with motor impairments of different origins (Authors et al., 2014; Cortinovis, Luraschi, Intini et al., 2011; Sznitman, Baruchb, Greeneb & Gelkopfb, 2018).

2.4. Bias

The bias of answer distortion due to retrospective recall was limited by discarding from analysis forms completed over 20 minutes after signal receipt, in order to, as retrospective reports are subject to memory biases, and several studies have detected only partial overlapping between retrospective ratings of mood and behaviors and real-time assessments (Schimmack, 2003). Moreover, a short time lag between signal and response ensures that ESM ratings validly reflect internal experiences and not individual's response styles (Schimmack, 2003) or social desirability (Hektner et al., 2007).

2.5. Statistical Methods

After collection, ESM data were preliminary screened for form completeness and response rate. The qualitative descriptions of ongoing activities were assigned numeric codes using extant manuals, and they were subsequently grouped in broader categories (Hektner et al., 2007). As for scaled variables, their standardized values (z-scores, M=0, SD=1) were calculated by subtracting each participant's mean value from the raw score, and then dividing the result by the participant's standard deviation. After standardization, each variable had as many z-scores as raw self-reports (except for possible missing values). This approach exploits the potential of ESM repeated assessments, as it allows researchers to explore the experience fluctuation over the week (Hektner et al., 2007), by using the individual's week average score of a variable as the within-person cut-off point to which to

compare daily moment-by-moment scores for that variable. At the inter-individual level, individuals' rating standardization (*z*-scores) provides a common metric to compare experience across participants.

Descriptive analyses first included the frequency distribution of the activity categories across groups. As concerns the relationship between experience and activity type, across *t*-tests were performed to assess whether the standardized values of the selected cognitive, affective and motivational variables associated with a given activity category significantly differed from average across autonomy groups and in the total sample (H1).

The role of autonomy level and activity type in predicting participants' experience (H2) and perceived skills and challenges (H3a and b) was investigated through a multilevel (ML) approach using variables' raw scores. ML analysis is suited for the ESM data structure (Raudenbush & Bryk, 2002), as it can adequately handle nested data with unequal numbers of observations across individuals and unequally spaced time intervals between observations. Moreover, it provides estimates of intra- and inter-individual variability, thus considering autocorrelations.

To verify (H2), the experiential dimensions selected for this study were taken as dependent variables in as many models, using autonomy level and activity type as predictors. Random intercepts for participants were included. To explore perceived skills and challenges across activities and autonomy groups (H3a and b), a unique bivariate three-level model was used, with responses concerning skills and challenges as dependent variable, and autonomy level, type of activity, and a dimension representing the relation between challenge and skills as predictors. The by-participants beep random intercepts were also included in this model, since both challenges and skills were reported in each ESM form.

Analyses were run within the R environment (R Development Core Team, 2009), through packages lme4 (Bates et al., 2014) and lmerTest (Kuznetsova et al., 2017). Random effects were considered to account for the non-independency of observations by- participants (Hox, 2002). Fixed

effects were evaluated through an analysis-of-variance approach. P-values were calculated using the Satterthwaite correction for degrees of freedom (Satterthwaite, 1946). After model fit, atypical outliers were removed using 2.5 SD of the residual errors as criterion, and the models were refitted (Baayen, 2008).

As concern the sample size, simulation studies by Hox and Maas (Hox and Maas, 2004; Maas and Hox, 2005) examined the accuracy of the standard errors for the fixed effects and variance components for different samples sizes at both levels and intraclass correlations. They found that sample sizes greater than 30 had a minimal impact on the accuracy of the standard error for the fixed effects. Sample sizes less than 30 led to standard errors that were too small, especially in the case of a large intraclass correlation.

3. RESULTS

3.1. Participants

Out of the 55 people originally contacted (21 women and 34 men), 50 joined the study. After the briefing session 7 declined participation, arguing that ESM would disrupt their daily routine; 5 dropped out after few sampling days due to difficulties in form completion; 3 interrupted participation due to acute health problems. The final sample thus comprised 35 participants, 16 women and 19 men, divided into three groups according to their BI score. Their demographic features are reported in Table 1. Consistent with epidemiological data (Martinuzzi, Montanaro, Vavla et al., 2016; Racis et al., 2014), most participants (60%) lived in Sardinia, 31.5% in North Italy, and 8.5 in Central and South Italy.

INSERT TABLE 1 ABOUT HERE

Participants in the LBI group were severely dependent (BI score in the 21-60 range). They moved in a wheelchair, had urinary incontinence, and needed help in daily activities. Participants in the MBI group (BI score within the 61-90 range) walked with crutches, had urinary continence, were

independent in eating and dressing, but needed help in performing complex movements like rolling on socks. Participants in the HBI group (BI score above 90) had urinary continence and were able to eat, dress and walk autonomously, though sometimes needing crutches.

No significant group differences emerged for demographic features. Overall, participants provided 1077 valid ESM forms. Consistent with studies involving persons with motor impairments, the response rate was 54.9% for the global sample, without differences across groups: LBI participants provided 352 forms (each 27.1 on average), MBI participants 486 forms (each 37.4 on average) and HBI participants 239 forms (each 26.6 on average).

3.2. Descriptive data: Activity distribution

Table 2 illustrates the distribution of daily activity categories. Productive activities comprised work, study, as well as manual activities performed by LBI participants in a day-care center. Leisure mainly included sedentary tasks such as crosswords, puzzles, video games, reading and watching TV; interactions involved family, friends and healthcare professionals; personal care comprised eating, resting, hygiene, medications and physiotherapy. The category "other" mainly comprised driving or traveling by car.

Across groups, the predominant activity category was leisure. Together with personal care it accounted for over 76% of the answers provided by LBI participants. Among MBI and HBI participants a more balanced answer distribution across categories was observed. Physiotherapy, included in personal care, was reported in only 8 forms, accounting for 3% of the total answers.

PLEASE INSERT TABLE 2 ABOUT HERE

3.3. Outcome data: Experience and activity type

The experience associated with each activity category (H1) was analyzed using the standardized values (z-scores) of the cognitive, emotional and motivational variables selected for this study; *t*-tests were preliminary performed to verify whether the experience associated with each activity category significantly differed from average across groups. Since no differences emerged, data were pooled together. Table 3 shows the experience associated to each activity category in the whole sample.

During productive activities participants were significantly more active than average (t=3.05), but also less happy (t=-2.19) and less wishing to do the activity (t=-3.87). During leisure control and wish to do the activity scored significantly higher than average (t=2.37 and t=2.81 respectively), while perceived activity stake scored significantly lower (t=-2.21). During interactions, most values were significantly above average: challenges (t=4.6), skills (t=2.24), happy (t=2.5), active (t=3.81), wish to do the activity (t=2.53) and perceived activity stake (t=3.24). A globally negative experience instead emerged during personal care, with significantly below average values of challenges (t=-3.27), concentration (t=-2.82), control (t=-3.5), and active (t=-3.46).

PLEASE INSERT TABLE 3 ABOUT HERE

3.4. **Main results (1): Experience variations according to activity type and autonomy level**

Variations in the experiential variables' values according to activity type and participants' autonomy level (H2) were identified through multi-level (ML) analysis. Productive activities were used as baseline for activity type, and LBI for autonomy level. Findings are illustrated in Table 4.

PLEASE INSERT TABLE 4 ABOUT HERE

No effect was observed for happiness, while the effect of activity type emerged for all the other variables. In particular, compared to the other activities personal care was associated to lower values of concentration (leisure vs. personal care C.I [0.88+/-0.11]; interactions vs. personal care: C.I. [0.63+/-0.42]; productive vs. personal care: C.I. [1.41+/-0.42]) and control (leisure vs. personal care C.I. [0.75+/-0.36]; interactions vs. personal care: C.I. [0.65+/-0.44]; productive vs. personal care: C.I.

[0.62+/-0.45]). Active scored lower in leisure and personal care (interactions vs. personal care: C.I. [1.17+/-0.45]; productive vs. personal care: C.I. [1.08+/-0.46]; interactions vs. leisure: C.I. [0.59+/-0.43]; productive vs. leisure: C.I. [0.46+/-0.46]); moreover, this variable scored lower during personal care than during leisure (C.I. [-1.18+/-0.96]). As for wish to do the activity, lower scores were detected during productive tasks (leisure vs. productive C.I. [1.6+/-0.6]; interactions vs. productive: C.I. [1.6+/-0.63]; personal care vs. productive: C.I. [1.34+/-0.6]).

A significant interaction between activity type and autonomy group emerged for perceived stake in the activity. Compared to the other two groups, HBI participants reported the lowest levels of perceived stake across activities (HBI vs. MBI: C.I. [2.55+/-1.56]; HBI vs. LBI: C.I. [3.56+/-1.58]). Variations in the relation between perceived stake and activity types instead emerged for the other groups. MBI participants perceived significantly higher stake levels during interactions (leisure vs. interactions C.I. [-1.14+/-0.81]; productive vs. interactions: C.I. [-1.16+/-0.9]; personal care vs. interactions: C.I. [-1.11+/-0.91]). Among LBI participants, stake levels were significantly higher during productive activities (leisure vs. productive: C.I. [-3.69+/-1.5]; interactions vs. productive: C.I. [-2.3+/-1.73]; personal care vs. productive: C.I. [-3.2+/-1.54]), as well as during interactions vis-à-vis leisure (CI: [1.39+/-1.13]).

3.5. **Main results (2). Bivariate model: Challenge/skill relationship according to activity type and autonomy level**

To verify H3a and H3b, a bivariate model was adopted. Autonomy level and activity type were simultaneously entered in the model as predictors, and challenges/skills were nested in participants. Figure 1 and Table 5 summarize the results.

Significant interactions emerged. Perceived skills were overall higher than challenges (C.I. [2.62+/-0.24]), and especially for HBI participants (C.I. [3.66+/-0.6]). Among MBI participants,

challenges showed a peak during interactions (leisure vs. interactions C.I. [-1.21+/-0.74]; productive vs. interactions: C.I. [-1.61+/-0.81]; personal care vs. interactions: C.I. [-2.24+/-0.83]), while skill values differed significantly between interactions and personal care (C.I. [0.82+/- 0.64]). LBI participants perceived higher challenges in productive activities (leisure vs. productive C.I. [-3.67+/-1.35]; interactions vs. productive: C.I. [-2.19+/-1.56]; personal care vs. productive: C.I. [-3.66+/-1.36]), whereas no significant difference across activities emerged for skills.

PLEASE INSERT FIGURE 1 AND TABLE 5 ABOUT HERE

4. DISCUSSION

4.1. Main results

The present study used a repeated sampling procedure to explore the daily experience of people with Hereditary Spastic Paraplegia (HSPs) with different autonomy levels. Data collected through Experience Sampling Method provided information on participants' real-time experience in their daily environment, and on their perception of daily activities as opportunities for positive engagement and optimal experiences. To the best of our knowledge, no studies have been conducted yet on this topic and through this methodology

Results showed that the main predictor of participants' experience were the perceived relevance and complexity of the ongoing activity, while the autonomy level played only a marginal role.

4.1.1. Daily activities, quality of experience and autonomy levels

After dividing participants in three groups according to their autonomy level, we first analyzed the distribution of their daily activities. Across groups, the most frequently reported activities were leisure and personal care. Leisure predominantly included passive and sedentary activities, such as watching TV and playing video games; support to this finding comes from studies conducted among persons with disabilities in other countries (Pagan-Rodriguez, 2014). Personal care activities mostly

comprised daily hygiene, eating, and resting, while physiotherapy and rehabilitation treatments accounted for very few self-reports, despite their well-established role in reducing spasticity and increasing muscle strength and cardiovascular function among persons with HSP (Fink, 2003).

Such a generalized lack of physical activity in leisure and personal care poses serious threats to the person's autonomy and quality of life over time, as it contributes to increased muscle stiffness and loss of muscle tone. Although an active lifestyle is recommended to people with physical disabilities, psychological and social barriers may interfere in the attainment of this goal; they include lack of infrastructures such as gyms or pedestrian areas; behavioral norms of the society; feelings of unease or shame; and social isolation (Delle Fave & Massimini, 2005b Richardson, Smith, & Papathomas, 2017). Evidence of these barriers emerged in the present study: Most participants lived in small towns with very limited facilities for physical activity; their community lifestyle did not include a regular sport practice. As concerns personal care, the negligible occurrence of physical treatments reported by the participants reflects the national healthcare provisions, according to which citizens requiring regular physiotherapy are offered eight weekly treatment sessions free of charge per year; longer treatment cycles are often very expensive for families, as well as problematic in terms of logistics.

In order to verify (H1), the standardized values of the experiential variables associated with each activity category were preliminarily compared across the three groups of participants. Since no significant differences emerged according to autonomy levels, the quality of experience associated to each activity category was analyzed on the pooled sample data. Different experiential profiles emerged in relation to the different activities, thus supporting our hypothesis.

More specifically, personal care was associated with a negative condition of cognitive disengagement and low challenges. The association of the repetitive and low-challenge personal care tasks with a globally negative experience was recurrently detected in ESM research, including studies involving persons with motor disabilities (Cortinovis et al., 2011; Authors et al., 2014).

An opposite, flow-like experiential profile was detected during interactions, characterized by positive values of affective and motivational variables, as well as significantly high values of both challenges and skills, as expected in flow experiences. This finding is in line with the literature underscoring the pivotal role of relationships in fostering perceived meaning and in supporting adaptive coping strategies among people with chronic diseases (Pakenham, 2008: Bassi, Falautano, Cilia et al., 2016).

A more nuanced experience profile emerged for the other two activity domains. During productive tasks participants perceived average challenge and skill levels, they were more active than average, but less happy and less willing to do the activity. This finding is consistent with the so-called work paradox: productive tasks support activation and focused attention, but not emotional and motivational involvement (Csikszentmihalyi & Lefevre, 1989). Nevertheless, work represents an important opportunity for cognitive and behavioral resource mobilization, and for flow experiences (for a review, see Fullagar & Delle Fave, 2017). Moreover, it promotes socialization and functional independence among people with disabilities (Schopp, Clark, Hagglund et al., 2007; Cortinovis et al., 2011).

During leisure – primarily including passive and sedentary activities - participants reported high control and wish to do the activity, but low stake of the activity. Passive leisure, being self-determined and pleasurable in the short term, may help buffer the negative effects of adversity and sustain coping efforts among people with chronic illness, but it is less effective in supporting commitment and skill mobilization over time (Wikström, Book & Jacobsson, 2006). To the contrary, structured and challenging activities such as sports, physical exercise, arts and crafts and they allow people with disabilities to experience flow, and at the same time to preserve functional autonomy, build new abilities or develop residual ones, and cultivate relationships (Delle Fave & Massimini, 2005b).

4.1.2. The relationship of activity type and autonomy level with the quality of experience

ML analyses allowed for the evaluation of the relationship between activity type, participants' autonomy level, and the cognitive, emotional and motivational components of the experience. Contrary to our hypothesis (H2), but consistent with other ESM studies (Bassi et al., 2012; Authors et al., 2014), autonomy level played a modest role, as no group differences emerged in the experience associated with each of the examined activities. The only exception was observed for the perceived stake of the activity, whose values were significantly higher among LBI participants, especially during productive activities. The manifold difficulties that daily tasks pose to people with low autonomy, but also their potential as opportunities for meaning-making and social inclusion (especially for productive activities and interactions) may help interpret this result.

Finally, a specific relationship pattern between perceived challenges and skill, activity type and autonomy level were expected (H3a andb), with challenges and skills attaining higher levels during productive activities and interactions than during leisure and personal care, and more autonomous participants reporting higher challenges during productive activities. Analyses highlighted that, across activities and groups, challenge levels were lower than skill ones; not surprisingly, this pattern was particularly evident for highly autonomous participants, who also perceived higher skills than the other two groups. These results suggest the ability of HBI participants to more effectively express and mobilize competences, especially in complex and refined tasks. Among MBI participants, a significant increase of challenges in relation to skills was observed during interactions; to interpret this finding itmay be useful to consider that these participants partially need other peoples' help to perform daily tasks, at the same time being sufficiently independent to actively search for social interactions, as opportunities to strengthen existing relationships as well as building new ones. Finally, among LBI participants perceived challenges hit the highest level in productive activities, often performed in a

daycare center: This result suggests the twofold role of these activities as opportunities for active engagement and social participation outside the domestic and family settings.

Overall, these results highlight the importance of tailoring rehabilitation programs on individuals' perceived challenges and abilities, in order to promote their active engagement in tasks representing both opportunities for skill improvement and socialization. More specifically, findings support our initial expectation about the relevance of productive activities and interactions in promoting complex and positive experiences such as flow, that can promote high performance, intrinsic rewards and the long-term development of competences and autonomy in the most diverse activity domains, from structured leisure (Freire, 2013) to work (Fullagar & Delle Fave, 2017).

4.2. Limitations, strengths and research directions

To the best of our knowledge, this is the first study investigating the daily experience of persons with neuromuscular diseases in real time and taking into account their autonomy level. At the same time, this study has several limitations. Data were collected cross-sectionally during a standard sampling week. The small sample size is not representative of the Italian HSPs population, thus preventing from result generalization.

At the methodological level ESM presents some disadvantages (Napa Scollon et al., 2003). Participants' self-selection bias and attrition (potentially emerging in any study) are relevant, due to the long sampling session and the onerous task of randomly filling in questionnaires during daily activities. Situational issues regard the possibility that individuals may not want to or could not respond to one or more signals in specific contexts (e.g. during religious rituals or sport competitions). However, the large majority of ESM participants reported that their week routine was comprehensively captured (Csikszentmihalyi & Larson, 1987), while only a minority complained about signals disrupting of their daily routine (22%; Hormuth, 1986).

Results from this study pave the way for new research avenues. Considering the well-established role of a positive relationship with caregivers in fostering well-being among people with neurological diseases (Trail, Nelson & Van, 2003; Pakenham, 2008;), the synchronous ESM based investigation of the experience reported by people with HSPs and their caregivers can shed light on convergences and divergences in perceived challenges and opportunities within a shared environment, offering health professionals useful information to design rehabilitation processes and to orient family coping strategies.

More generally, experience sampling studies can offer a better understanding of the daily experience of people diagnosed with chronic or progressive disorders. Despite related challenges, the fine-grained picture of daily experiences provided by ESM may help identify unmet needs and hidden resources, that can orient person-tailored interventions aimed at supporting the adaptive management of daily life and the optimal developmental trajectory of persons with disabilities.

5 CONCLUSIONS

Results from this study suggest that, regardless of their autonomy level, persons with HSPs should be supported in finding more challenging opportunities for action in daily life, in order to attain engaging and optimal experiences. The exercise of motor functions and the cultivation of vicarious ones should be promoted through physiotherapy programs and physical activity practice. Job placement should be favored, as productive activities can foster engaging experiences, as well as represent opportunities for personal growth and social inclusion. Finally, social interactions should be facilitated through engagement in family and community activities.

References

Authors et al. (2014).

Baayen, R. H. (2008). *Analyzing Linguistic Data: A Practical Introduction to Statistics using R*. Cambridge University Press.

Bassi M., Delle Fave A. (2012). Optimal experience and self-determination at school: Joining perspectives. *Motivation and Emotion, 36,* 425-438.

Bassi, M., Falautano M., Cilia S., Goretti B., Grobberio M., Pattini M., Pietrolongo E., Viterbo R.G., Amato M.P., Benin M., Lugaresi A., Minacapelli E., Montanari E., Patti F., Trojano M., Delle Fave A. (2016). Illness perception and well-being among persons with multiple sclerosis and their caregivers. *Journal of Clinical Psychology in Medical Settings, 23,* 33-52. DOI 10.1007/s10880-015-9425-8.

Bassi M., Ferrario N., Ba G., Delle Fave A., & Viganò C. (2012). Quality of experience during psychosocial rehabilitation: a real-time investigation with Experience Sampling Method. *Psychiatric Rehabilitation Journal, 35,* 447–453. http://dx.doi.org/10.1037/h0094578.

Bates, D., Mächler, M., Bolker, B., & Walker, S. (2014). Fitting linear mixed-effects models using lme4. *arXiv preprint arXiv:1406.5823*.

Benchimol, E.I., Smeeth, L., Guttmann, A., Harron, K., Moher, D., Petersen, I., Sorensen,H.T., von Elm, E., Langan, S.M., Committee, R.W. (2015). The REporting of studies Conducted using Observational Routinely-collected health Data (RECORD) statement. *PLoS Med, 12 (10)*: e1001885. DOI:10.1371/journal.pmed.1001885.

Blackstone, C., O'Kane, CJ. & Reid, E. (2011). Hereditary Spastic Paraplegia: Membrane Traffic and Motor Pathway. *Nature Reviews Neuroscience, 12,* 31–42. DOI: 10.1038/nrn2946.

Conner T.S., Tennen H., Fleeson W. & Feldman Barrett L. (2009). Experience sampling methods: A modern idiographic approach to personality research. *Social and Personality Psychology Compass, 3,* 1-22.

Cortinovis, I., Intini, E., Luraschi, S., Sessa, M., & Delle Fave, A. (2011). The daily experience of people with Achondroplasia. *Applied Psychology: Health and Well-being, 3(2)*, 2017-227. DOI: 10.1111/j.1758-0854.2010.01046.x.

Csikszentmihalyi, M. (1975/2000). *Beyond Boredom and Anxiety*. San Francisco: Jossey-Bass.

Csikszentmihalyi, M., & LeFevre, J. (1989). Optimal experience in work and leisure. *Journal of Personality and Social Psychology, 56*, 815–822.

Delle Fave, A. & Massimini, F. (2005a). The investigation of optimal experience and apathy: developmental and psychosocial implications. *European Psychologist, 10*, 264-274. https://doi.org/10.1027/1016-9040.10.4.264.

Delle Fave, A. & Massimini, F. (2005b). The relevance of subjective well-being to social policies: Optimal experience and tailored intervention. In: F. Huppert, N. Baylis & B. Keverne (Eds.) *The science of well-being*, (pp. 379–402). Oxford, Oxford University Press.

Delle Fave, A., Massimini, F., & Bassi, M. (2011). *Psychological selection and optimal experience across cultures: Social empowerment through personal growth*. Dordrecht, NL: Springer.

Fink, J.K. (2003). Advances in the hereditary spastic paraplegias. *Experimental Neurology, 184,* 106–10. https://doi.org/10.1016/j.expneurol.2003.08.005.

Folkman, S., & Moskowitz, J.T. (2000). Stress, positive emotions and coping. *Current directions in Psychological Science, 9,* 115–118.

Freire, T. (2013, Ed.), *Positive leisure science: From subjective experience to social contexts*. Dordrecht, NL: Springer.

Fullagar, C., & Delle Fave, A. (2017 Eds.). *Flow at work: Measurement and implications*. NY: Taylor & Francis.

Fullagar, C. J., Knight, P. A., & Sovern, H. S. (2013). Challenge/skill balance, flow, and performance anxiety. *Applied Psychology: An International Review, 62,* 236–259. DOI: http://dx.doi.org/10.1111/j.1464-0597.2012.00494.x.

Hektner, J. M., Schmidt, J. A., & Csikszentmihalyi, M. (2007). *Experience Sampling Method: measuring the quality of everyday life*, Sage Publication.

Hox, J. J. (2002). *Multilevel Analysis: Techniques and applications* (2nd edition). Routledge, Taylor & Francis, New York.

Hox, J. & Maas, C.J.M. (2004). Sufficient Sample Size for Multilevel Modelling. *Methodology European Journal of Research Methodoology for the Behavioral and Social Sciences*. DOI: 10.1027/1614-2241.1.3.85.

Klebe, S., Stevanin, G., & Depienne C. (2015). Clinical and genetic heterogeneity in hereditary spastic paraplegias: From SPG1 to SPG72 and still counting. *Review in Neurology, 171*, 505–530. DOI: 10.1016/j.neurol.2015.02.017.

Kuznetsova, A., Brockhoff, P. B., & Christensen, R. H. B. (2017). lmerTest package: tests in linear mixed effects models. *Journal of Statistical Software, 82(13*).

Lo Giudice, T., Lombardi, F., Santorelli, FM., Kawarai T., & Orlacchio, A. (2014). Hereditary Spastic Paraplegia: clinical-genetic characteristics and evolving molecular mechanism. *Experimental Neurology, 261*, 518–539. DOI: 10.1016/j.expneurol.2014.06.011.

Mass, C.J.M. & Hox, J. (2005). Sufficient Sample Size for Multilevel Modelling. Methodology 2005; Vol. 1(3):86–92. DOI: 10.1027/1614-1881.1.3.86.

Mahoney, F.I., Barthel, D. (1965). Functional evaluation: The Barthel Index. *Maryland State Medical Journal, 14*, 56–61.

Martinuzzi, A., Montanaro, D., Vavla, M., Paparella, G., Bonanni, P., & Musumeci O. et al., (2016). Clinical and Paraclinical Indicators of Motor System Impairment in Hereditary Spastic Paraplegia: A Pilot Study. *PLOS ONE,* 1-19. DOI: 10.1371/journal.pone.0153283.

Massimini, F., & Delle Fave, A. (2000). Individual development in a bio-cultural perspective. *American Psychologist, 55,* 24–33. http://dx.doi.org/10.1037/0003-066X.55.1.24

McDermott, C, White, K, Bushby, K & Shaw, P. (2000). Hereditary spastic paraparesis: a review of new developments. *Journal of Neurological and Neurosurgery Psychiatry, 69*,150–160.

Mill, A., Kööts-AusmeesL., Allik, J., & Realo, A. (2018) The role of co-occurring emotions and personality traits in anger expression. Frontiers in Psychology, doi:10.3389/fpsyg.2018.00123.

Napa Scollon C., Kim-Prieto C., Diener E. (2003). Experience sampling: Promises and pitfalls, strengths and weaknesses. *Journal of Happiness Studies, 4,* 5-34.

Pakenham, K.I. (2008). Making sense of illness or disability: The nature of sense making in Multiple Sclerosis (MS). *Journal of Health Psychology, 13(1),* 93–105. DOI:10.1177/1359105307084315.

Pagan-Rodriguez, R. (2014). How do disabled individuals spend their leisure time? *Disability and Health Journal, 7,* 196–205. DOI: 10.1016/j.dhjo.2014.01.001.

Pagnini, F. (2013). Psychological well-being and quality of life in amyotrophic lateral sclerosis: A review. *International Journal of Psychology, 48(3),* 194-205. DOI: 10.1080/00207594.2012.691977.

Piccininni, M., Falsini, C. & Pizzi, A. (2004). Quality of life in hereditary neuromuscular diseases. *Acta Neurologica Scandinavica, 109*, 113–119. DOI: 10.1046/j.1600-0404.2003.00185.x.

Putnam, M., Greenen, S. & Powers, L. (2003). Health and wellness: People with disabilities discuss barriers and facilitators to well-being. *Journal of Rehabilitation, 69(1),* 37–45.

R Development Core Team (2009). R: A Language and Environment for Statistical Computing [Computer Software]. Avilable at: http://www.R-project.org

Racis, L., Tessa, A., Di Fabio, R., Storti, E., Agnetti, V., Casali, C., Santorelli, F. M., & Pugliatti, M. (2014). The high prevalence of hereditary spastic paraplegia in Sardinia, insular Italy. *Journal of Neurology, 261,* 52–59. DOI: 10.1007/s00415-013-7151-4.

Raudenbush, S.W., & Bryk, A. S. (2002). *Hierarchical Linear Models: Applications and data analysis methods* (2nd ed.). Thousand Oaks, CA: Sage Publications, Inc.

Richardson, E.V., Smith, B., & Papathomas, A. (2017). Disability and the gym: experiences, barriers and facilitators of gym use for individuals with physical disabilities. *Disability and Rehabilitation, 39,* 1950–1957. DOI: 10.1080/09638288.2016.1213893.

Ruano, L., Melo, C., Silva MC., & Coutinho, P. (2014). The global epidemiology of hereditary spastic paraplegia: a systematic review of prevalence studies. *Neuroepidemiology, 42,* 174–83. DOI: 10.1159/000358801.

Satterthwaite, F. E. (1946). An approximate distribution of estimates of variance components. *Biometrics bulletin, 2(6),* 110-114.

Schimmack, U. (2003). Affect measurement in experience sampling research, *Journal of Happiness Studies, 4,* 79-106.

Schopp, L. H., Clark, M. J., Hagglund, K. J., Sherman, A. K., Stout, B. J., Gray, D. B. et al., (2007). Life activities among individuals with Spinal Cord Injury living in the community: Perceived choice and perceived Barriers. *Rehabilitation Psychology, 52,* 82–88.

Shah S., Vanclay F., & Cooper B. (1989). Improving the sensitivity of the Barthel Index for stroke rehabilitation. *Journal of Clinical Epidemiology, 42,* 703–9.

Simmons, Z. Bremer, B. A. Robbins, R. A. Walsh S. M., Fischer, S. (2000). Quality of life in ALS depends on factors other than strength and physical function. *Neurology, 55(3),* 388–392.

Sznitman, S., Baruc, Y.B., Green, T., & Gelkopf, N. (2018). The association between physical pain and cannabis use in daily life: An experience sampling method. *Drug and Alcohol Dependence*, https://doi.org/10.1016/j.drugalcdep.2018.07.014.

Taruscio D., Kodra, Y., Ferrari, G., Vittozzi, L. and the National Rare Diseases Registry Collaborating Group (2014). The Italian National Rare Diseases Registry. *Blood Transfusion*, 12, Suppl 3: s606-13 DOI 10.2450/2014.0064–14s.

Trail, M., Nelson, N. D. e Van, J. N. (2003). A study comparing patients with amyotrophic lateral sclerosis and their caregivers on measures of quality of life, depression and their attitudes toward treatment options. *Journal of the Neurological Sciences, 29*, 79–85.

Trull T.J. & Ebner-Priemer U.W. (2009). Using experience sampling methods/ecological momentary assessment (ESM/EMA) in clinical assessment and clinical research: Introduction to the Special Section. *Psychological Assessment, 21,* 457-462.

Wechsler, D. (1981). *WAIS-R. Scala d'Intelligenza Wechsler per Adulti-Riveduta*. Tr. it. O.S. Organizzazioni Speciali, Firenze 1997.

Wikström, I., Book, C., Jacobsson, L. T. H. (2006). Difficulties in performing leisure activities among persons with newly diagnosed rheumatoid arthritis: a prospective, controlled study. *Rheumatology, 45*, 1162–1166. DOI:10.1093/rheumatology/kel080.

World Health Organization. (2001). *International Classification of Functioning, Disability and Health (ICF)*. Geneva: WHO.

Tables

Table 1. Participants' demographic and clinical characteristics

Table 2. Distribution of daily activity categories across groups

Table 3. Average experience in the main activity categories

Table 4. Multi-level analysis: experiential variables' fluctuation with autonomy level and activity type as predictors

Table 5. Bi-variate multi-level analysis: challenge and skill fluctuation with autonomy level, activity type and challenge/skill relation as predictors

Figures

Figure 1. Predicted fluctuation of challenges and skills across autonomy groups and activity types

Table 1.

	LBI[a] **(N=13)**	**MBI**[b] **(N=13)**	**HBI**[c] **(N=9)**
Age[d]	47.8 (21.3)	48.6 (7.3)	43.9 (13.9)
Gender (% women)	53.84	30.77	55.56
Education (%)			
Elementary or middle school	69.2	56.1	100
High school/University	30.8	43.9	-
Employment (%)			
Retirement pension	46.1	53.8	12.5
Disability pension	23.1	-	12.5
Daycare center for people with disabilities	15.4	-	-
Workers	-	30.8	-
Housewives	7.6	15.4	75
Civil status			
Single/separated/divorced	61.5	7.6	55.6
Married/cohabiting	38.5	92.4	44.4
Barthel Index (BI)[d]	35 (12.8)	77.7 (10.1)	96.1 (2.2)
SPG type (%)			
Autosomal dominant forms (SPG4)	7.6	53.9	77.8
Autosomal recessive forms (SPG7-SPG11-SPG30)	53.9	15.4	11.1
Genetically unidentified forms	38.5	30.7	11.1
Mild/moderate mental retardation (%)	53.9	-	-

Note. N=Number of participants; [a]= low autonomy in daily activities; [b]= moderate autonomy in daily activities [c]= high autonomy in daily activities; [d]=mean value and (sd) are reported

Table 2.

Activity categories	**LBI**	**MBI**	**HBI**
	%	**%**	**%**
Productive activities	6,5	21,2	20,5
Leisure	43,2	36,4	28,9
Interactions	10,5	16,9	21,8
Personal care	32,7	19,1	24,3
Others	7,1	6,4	4,5
N. answers	**352**	**486**	**239**
N participants	**13**	**13**	**9**

Note: N= No. of participants

Table 3.

	Productive activities		**Leisure**		**Interactions**		**Personal care**	
	N[a] =35		**N[a] =35**		**N[a] =35**		**N[a] =35**	
	M	**sd**	**M**	**sd**	**M**	**sd**	**M**	**sd**
Challenges	.09	.92	-.04	.97	.36***	1.02	-.19**	.97
Skills	-.09	1.01	.04	.89	.16*	.96	-.10	1.1
Concentration	.05	.8	.06	.93	.13	.93	-.19*	1.1
In control	.04	.9	.11*	.89	.07	1.09	-.23***	1.05
Happy	-.15*	.87	.03	.94	.19*	.98	-.06	1.04
Active	.21**	.88	-.03	.96	.26***	.88	-.24*	1.09
Wish to do the act.	-.31***	1.05	.12**	.85	.16*	.83	.01	.98
Stake of the activity	.03	.88	-.11*	.92	.25**	1.01	-.03	1
N. answers	**175**		**390**		**171**		**266**	

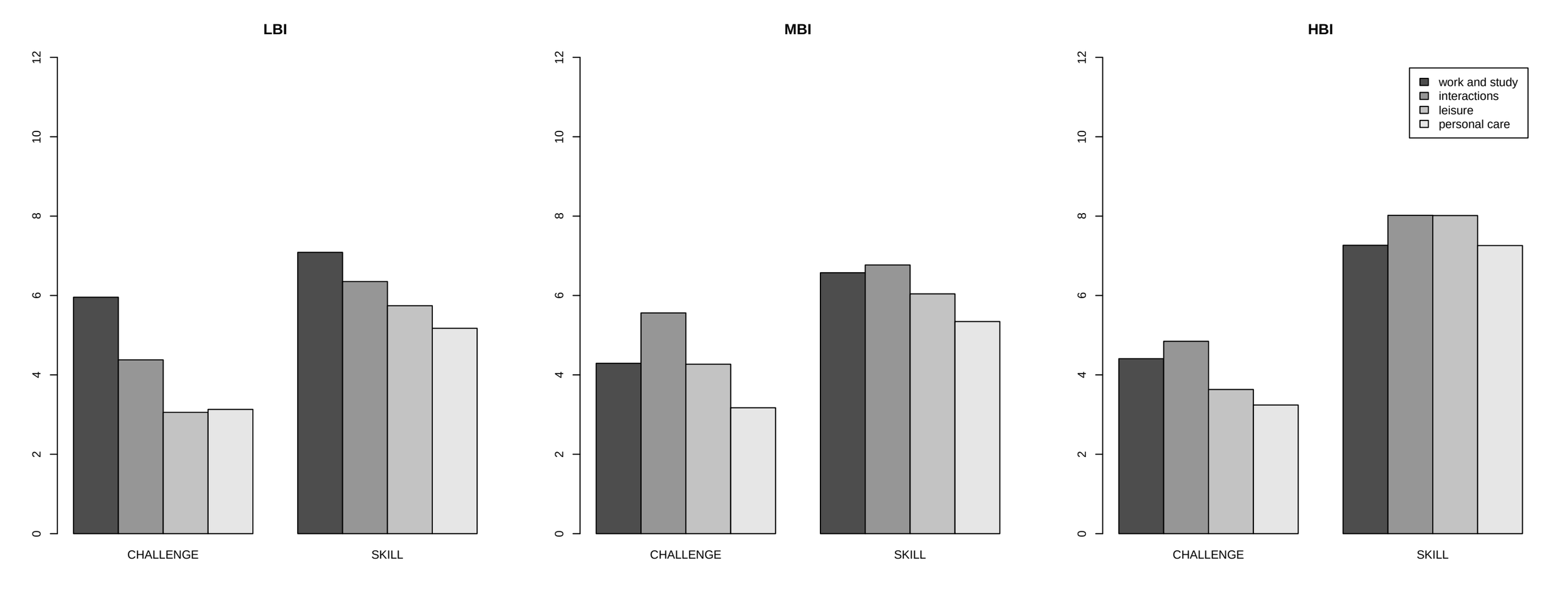
*=p< 0.05 **=p< 0.01 ***=p< 0.001 N[a] = number of participants.

Table 4.

	Fixed effect estimates	SE	T	p
Concentration				
Intercept	6.268	0.372	16.84	.0001
Leisure	0.032	0.201	0.16	.8712
Interactions	0.225	0.229	0.98	.3261
Personal care	-0.405	0.215	-1.87	.0608
Control				
Intercept	6.613	0.417	15.83	.0001
Leisure	0.105	0.192	0.54	.5829
Interactions	0.129	0.219	0.58	.5560
Personal care	-0.453	0.206	-2.19	.0287
Happy				
Intercept	5.050	0.401	12.57	.0001
Leisure	0.461	0.209	2.20	.0279
Interactions	0.645	0.239	2.69	.0071
Personal care	0.172	0.226	0.76	.4453
Active				
Intercept	5.767	0.348	16.52	.0001
Leisure	-0.488	0.219	-2.22	.0261
Interactions	0.092	0.250	0.36	.7129
Personal care	-1.079	0.236	-4.55	.0001

Table 5.

	F	df Numerator	df Denominator	p
Autonomy	0.39	2	32.88	.68
Activity	16.75	3	1886.41	<.001***
Chall/skill relation	34.94	1	33.71	<.001***
Activity*Autonomy	3.85	6	1884.27	<.001***
Chall/skill relation*Autonomy	2.99	2	33.69	.06
Activity*Chall/skill relation	7.87	3	1852.61	<.001***
Autonomy*Activity*Chall/skill relation	3.79	6	1851.76	<.001***


LBI
MBI
HBI
0
2
4
6
8
10
12
CHALLENGE
SKILL
work and study
interactions
leisure
personal care