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The clinical utility of the Social Responsiveness Scale and Social Communication Questionnaire in tuberous sclerosis complex

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ABSTRACT

Tuberous sclerosis complex (TSC) is often associated with epilepsy, mental retardation, and autism spectrum disorders (ASDs). Thus, screening for ASDs is important when evaluating these individuals. We examined the utility of the Social Responsiveness Scale (SRS) and Social Communication Questionnaire (SCQ), two measures for screening for ASDs, in a TSC population. Twenty-one children were evaluated, with 52.4% classified as having ASDs on the SRS and 42.9% classified as such on the SCQ. Number of antiepileptic drugs significantly correlated with SRS Total score, as did level of intellectual functioning. Evidence for convergent validity was obtained between the SRS and SCQ Total scores ($r = 0.605$). Moreover, all SRS subscales correlated with SCQ Total score ($r > 0.400$). All SCQ subscales except for Communication correlated with SRS total. Overall, the results demonstrate that these questionnaires appear to be effective screens for ASDs in a TSC population and are measuring similar constructs.

1. Introduction

First described in the 1800s, tuberous sclerosis complex (TSC) is a genetic disorder associated with mutation of either the TSC1 gene (9q34) or the TSC2 gene (16p13.3) [1,2]. TSC is inherited as an autosomal dominant trait, but 60 to 70% of cases are sporadic and may represent new mutations [3–5]. The prevalence of TSC ranges from 1 in 6000 live births [5] to 1 in 10,000 live births [6]. One study estimated that the total population prevalence was between 7 and 12 cases per 100,000, with more than half of cases remaining undetected [7].

The presentation of TSC is quite variable. Patients with TSC often have tumors in several organ systems such as the skin, eyes, kidneys, and brain. Cerebral involvement is very common, occurring in approximately 90% of cases [8]. Tubers are most frequently located in the frontal, parietal, and temporal lobes, with degree of cerebral involvement (i.e., tuber burden) varying widely across patients [9]. Epilepsy is a common feature of TSC, occurring in approximately 75 to 92% of all cases, with 50% initially presenting with infantile spasms [10,11].

Furthermore, TSC is often associated with mental retardation, learning disabilities, and autism spectrum disorders (ASDs). The prevalence of mental retardation in TSC has been estimated to range from 44 to 60% [12,13]. Tuber count is associated with cognitive outcomes, with more tubers associated with high risk of intellectual disability [14,15]. Those with mutations on TSC2 are more likely to have mental retardation [16]. Another risk factor for intellectual impairment is the occurrence of infantile spasms, as early seizure onset is associated with delayed development and intellectual disabilities in general [17–19]. However, Raznahan and colleagues reported that the number of tubers in an individual is more strongly associated with intellectual disability than is a history of infantile spasms [20].

Intellectual disability and infantile spasms are associated with ASDs in TSC [21,22]. ASDs occur in approximately 25 to 50% of people with TSC [23] and are characterized by deficits in reciprocal social interactions, communication abilities, and stereotyped behaviors, which are present early in development [24]. ASDs, like most behavioral syndromes, are an etiologically heterogeneous disorder, which can often complicate diagnosis [25]. One study by Jeste and colleagues found that children with TSC and ASDs were more cognitively impaired than those without autism and demonstrated no differences between social and communication scores on the Autism Diagnostic Observation Schedule [26].

Given the high rate of ASDs in children with TSC, there is a need for screening measures of social and emotional functioning that are appropriate for this population. The "gold" standard for ASD diagnosis is currently the Autism Diagnostic Interview—Revised (ADI-R) [27,28], which can take 2 hours or longer to complete, and the Autism Diagnostic

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Observation Schedule (ADOS) [29]. These measures can be onerous to administer and are often not practical in clinical settings; consequently, many clinicians do not administer such assessments as a routine part of their evaluation. Rapid detection of ASDs permits early diagnosis and treatment, which will maximize the prognosis for the patient [30,31].

The Social Responsiveness Scale (SRS) [32] and the Social Communication Questionnaire (SCQ) [33] are two commonly administered screening instruments for ASDs. One advantage of the SCQ is that it was developed in tandem with the ADI-R, and thus, the ADI-R and the SCQ show a strong association. The SRS provides a good index of the severity of autistic social impairment, and previous studies have shown good convergent validity between the SRS and SCQ in populations of children with special educational needs (with and without ASDs), with correlations as high as 0.68 [34].

To our knowledge, no published study has compared the psychometric properties of the SRS and SCQ in a population of children with TSC. Though it is clear that many children with TSC meet criteria for ASDs, given the heterogeneous presentation of ASDs, it is not well understood if the presentation of ASDs in TSC differs meaningfully from that of idiopathic ASDs. Accordingly, the intercorrelations of these measures may differ within a TSC population.

An increased understanding of the psychometric properties of the SRS and SCQ in children with TSC will improve the utility of these measures in both clinical and research settings. The primary aim of the present investigation was to evaluate the convergent validity of these measures in individuals with TSC. A secondary aim was to explore the relationships among the subscales within each instrument, and a tertiary aim was to assess the relationships of the SRS and SCQ with epilepsy and cognitive variables commonly observed in children with TSC.

2. Methods

2.1. Participants

Participants included 21 parents of children with TSC who were recruited as part of a larger, ongoing study at the Comprehensive Epilepsy Center of New York University Medical Center. The children were between 4 and 18 years of age, with a diagnosis of TSC made in accordance with guidelines agreed on at the Tuberous Sclerosis Consensus Conference [35]. All parents and/or caregivers of patients were English-speaking. They signed informed consent forms at the time of data collection, and the children/adolescents also signed assent. All study procedures were approved by the institutional review board at the New York University Langone Medical Center in New York. A chart review was conducted to confirm diagnosis and gather information on intellectual functioning and seizure history. Parents of patients completed the SRS and SCQ.

2.2. Measures

2.2.1. Epilepsy variables

Epilepsy-related variables were obtained by chart review and/or interview with parents pertaining to the patient's seizure history. These included occurrence of infantile spasms, age at onset of seizures, and number of antiepileptic drugs (AEDs) currently prescribed.

2.2.2. Cognitive variables

Full Scale Intelligence Quotient (FSIQ) was assessed with the Wechsler Abbreviated Scale of Intelligence, Wechsler Intelligence Scale for Children IV, or Wechsler Adult Intelligence Scale III. Of the 21 children evaluated, 6 had estimated FSIQs that fell below the floor of these instruments (i.e., valid IQ scores were unable to be generated). For these individuals intellectual functioning was estimated with the Adaptive Behavior Composite score of the Vineland Adaptive Behavior Scales, Second Edition [36]. To classify the participants in terms of intellectual functioning, the 21 participants were placed into seven categories based on IQ scores, if available, or Adaptive Behavior Composite scores. Categorical classification criterion was based on the DSM-IV-TR for intellectual disability. The following labels were applied: "average intellectual functioning" (90–110), "low average intellectual functioning" (80–89), "borderline intellectual functioning" (70–79), "mild intellectual disability" (55–69), "moderate intellectual disability" (41–54), "severe intellectual disability" (40–26), and "profound intellectual disability" (<25).

2.2.3. Social Responsiveness Scale

The SRS is a 65-item questionnaire measuring autistic traits in children aged 4 to 18. Five domains are assessed: social awareness, social information processing, capacity for reciprocal social communication, social anxiety/avoidance, and autistic preoccupations and mannerisms. Each item is scored on a Likert scale ranging from 1 (not true) to 4 (almost always true). Raw scores are converted to *T* scores and a SRS Total *T* score of ≥ 70 is highly suggestive of an ASD. Excellent psychometric properties have been previously established for the SRS for use in children and adolescents [37,38]. Internal consistency is high for both male and female participants (Cronbach's $\alpha > 0.90$) and the instrument has shown temporal stability (test–retest reliability at 17 months: $r = 0.85$ for males and 0.77 for females). Interrater reliability is also high between mothers and fathers (0.91). The SRS discriminates well between children with ASDs and other psychiatric conditions [36]. Moreover, moderately strong associations were found between the SRS and the ADI-R, with correlation coefficients exceeding 0.52 across all subscales [39].

2.2.4. Social Communication Questionnaire

The SCQ is a 40-item (yes/no response format) questionnaire that evaluates communication skills and social functioning, both historically and currently. There are two different versions of the SCQ, the SCQ AutoScore™ Form: Lifetime and the SCQ AutoScore™ Form: Current. This study used the Lifetime version, which assesses behaviors that occur over the patient's life, with questions 20 through 40 focusing on behaviors occurring between the ages of 4 and 5. Total scores can range from 0 to 39 (the first question is a language screening item that is not included in the final score), and a total SCQ raw score of ≥ 15 is highly suggestive of ASD. Three theoretically derived subscales corresponding to ADI-R domains have been created, but not extensively researched. These include the Reciprocal Social Interaction subscale, Communication subscale, and the Restricted, Repetitive, and Stereotyped Patterns of Behavior subscale. The SCQ Total score shows strong relations with the ADI-R ($r = 0.71$) [40]. Moreover, internal consistency for the Total scale is high (Cronbach's $\alpha = 0.90$). Receiver operating curve analysis suggests an optimal cutoff score of 15 yielding a sensitivity of 0.85 and a specificity of 0.75.

2.3. Statistical analysis

Pearson's correlation coefficients were used to examine the bivariate relationships among the SRS and SCQ subscales, as well as correlations between these two instruments. Moreover, Pearson's correlation coefficients were used to examine the degree of association between the epilepsy variables and the SRS Total *T* score, as well as the SCQ Total score. Nonparametric statistics (i.e., Spearman's ρ) were used to assess the relationships between the categories of intellectual functioning and the SRS Total T score, as well as the SCQ Total score.

3. Results

Twenty-one parents and/or caregivers of children with TSC completed study instruments. Patients with TSC included 12 males and 9 females ranging in age from 5 to 18 (mean = 10.14, SD = 4.33). Twenty children had comorbid epilepsy. Of those with epilepsy, 55% had a history of infantile spasms (see Table 1). Bivariate relationships