Measuring Functional Status in Children With Genetic Impairments

MICHAEL E. MSALL* AND MICHELLE R. TREMONT

One of the consequences of genetic impairments in early childhood is their long-term effect on children's developmental skills in communication, learning, and adaptive behaviors. Functional assessment provides families and clinicians with a common language for describing a child's strengths and limitations in self-care (feeding, dressing, grooming, bathing, continence), mobility, and communication/social cognition. The National Center for Medical Rehabilitation Research described a model of disablement that includes five dimensions: pathophysiology, impairment, functional limitations, disability, and societal limitations. Using this framework, along with the Functional Independence Measure for children, the WeeFIM, we describe functional strengths and challenges in children with Down syndrome, spina bifida, congenital limb anomalies, congenital heart disease, urea cycle disorders, severe multiple developmental disabilities, and Di-George malformation sequence. We also briefly describe several pediatric functional/adaptive assessment instruments used by developmental professionals (Battelle Developmental Inventory, Vineland Adaptive Behavior Scales, Amount of Assistance Questionnaire). By tracking functional status, health professionals can prioritize secondary and tertiary prevention strategies that optimize self-care, mobility, communication, and learning. When functional limitations interfere with the acquisition of these essential skills, family and community support programs can be maximized. Am. J. Med. Genet. (Semin. Med. Genet.) 89:62–74, 1999. © 1999 Wiley-Liss, Inc.

KEY WORDS: functional assessment; genetic disabilities; developmental outcomes; Down syndrome; spina bifida; limb malformations; family supports; inborn errors of metabolism

INTRODUCTION

One of the key needs of families with children with genetic impairments is to understand the long-term impact of the disorder on the child's development. This cannot be viewed as a single measurement process at one point in time. Although the average intelligence quotient (IQ) of children with Down syndrome at kindergarten entry is two to three standard deviations below that of peers, this assessment will not document the child's ability to learn letters and numbers and apply these concepts to reading and mathematics. This IQ score does not tell professionals if the child runs, dresses, maintains continence, communicates basic needs, or plays games with friends. Thus, many activities most familiar to parents and professionals cannot be described by traditional psycho-educational scores.

Over the past decade a broader view of functional assessment in essential activities of self-care, mobility, communication, and social learning has emerged simultaneously with enhanced policy mandates established by Public Laws (PL) in early intervention (PL99-457), special education supports (PL94-142), employment opportunities (PL101-336), and disability assessment for Supplemental Security Income (SSI) [Perrin and Stein, 1991; Perrin et al., 1999]. The purpose of this review is to describe the use of functional assessment in children with genetic impairments. Functional assessment will be viewed as a process of describing a child’s strengths and challenges in the context of essential activities that occur within a child’s everyday environment.

BACKGROUND

More than 60 years ago, Doll understood that in order to optimize vocational outcomes and independent living...
everyday activities was essential [Doll, 1935]. These social competencies included motor milestones, hand skills, toileting, eating, dressing, locomotion, doing chores, communication, self-direction, and socialization [Doll, 1965]. Simultaneously, Denver and Brown [1945] introduced a measure for physical activities of daily living in order to assess skills required for community participation and employment training by adolescents and adults with chronic physical disability. Dimensions of this rating scale included locomotion, self-care, toileting, eating, and hand activities. Deaver’s significant contribution was the finding that time spent on a task in essential activities

| TABLE I. NCMRR Illustrations Model of Disablement and AAMR Family Support Models* |
|---------------------------------|---------------------------------|---------------------------------|---------------------------------|
| Definitions | Five-year-old boy with Down syndrome | Five-year-old boy with spina bifida | Five-year-old girl with DiGeorge sequence |
| Pathophysiology | Molecular, biochemical, or cellular mechanisms | Trisomy 21 | First trimester disorder of neural tube formation | Deletion of chromosome 22 |
| Impairment | Loss of structure or function at an organ level | Communicative and cognitive dysfunction | L3-L4 spinal cord dysfunction with neurogenic bowel and bladder and shunted hydrocephalus; weak hip extensors, hip abductors, and quadriceps | >90 db sensorineural hearing loss, tetralogy of Fallot, immune dysfunction |
| Functional limitation | Inability of a person to perform an activity normal for peers | Difficulty with receptive and expressive language, complex, self-care skills, unsupervised play, and problem solving | Difficulty with stairs and walking community distances of 1 block; difficulties with fasteners | Inability to hear sounds of language or to communicate verbally in sentences, difficulty with fine motor skills |
| Disability | Expression of functional limitation in a societal context of social roles typical for nondisabled peers | Early intervention resources for language, adaptive skills, socialization, and appropriate behaviors; family respite services | Intermittent clean catheterization, orthoses, and walker for ambulation and balance | Educational curricula emphasizing total communication, kindergarten readiness, and inclusion |
| Societal limitations | Legal, attitudinal, and cultural barriers affecting community participation | Easily accessible family supports for behavior challenges, creative curricula that optimize strengths and functional academics | Failure to allow for alternative efficient mobility, attitudes toward crawling at home, continency stigma | Too much focus on cochlea, social isolation of child, testimonials about educational philosophies |
| Family supports: AAMR definitions | Intermittent: Episodic or short-term developmental supports for life transitions | Down syndrome parent support group, exceptional parent newsletter, language software | Spina bifida parent support group; exceptional parent newsletter; small group efforts to enhance transfers, continency, and activities of daily living | Medical home supplemental Social Security Income, hearing-impaired parent support group |

*NCMRR, National Center for Medical Rehabilitation Research; AAMR, American Association of Mental Retardation.
was a key component for independent living.

**DISABILITY ASSESSMENT**

In the 1990s, the Institute of Medicine, the National Center for Medical Rehabilitation Research (NCMRR), the American Association on Mental Retardation, and the American Academy of Cerebral Palsy and Development Medicine promulgated models for conceptualizing disability [Butler et al., 1999]. Table I summarizes key concepts for defining levels of analysis for assessing disability and degrees of family supports (intermittent, limited, extensive, pervasive) from the American Association on Mental Retardation classification system [Luckasson et al., 1999]. Table I also illustrates applications of this model by comparing Down syndrome, spina bifida, and DiGeorge malformation sequence.

**PEDIATRIC FUNCTIONAL ASSESSMENT**

A multidisciplinary team of health, education, and rehabilitation professionals developed the Functional Independence Measure for children (WeeFIM®) to measure performance in self-care, continence, mobility, locomotion, communication, and social cognition [Msall et al., 1993]. The WeeFIM covers 18 items that describe seven levels of independence [Msall et al., 1990]. These efforts built on the pioneering efforts of Granger and colleagues to develop an essential database for describing the outcomes of medical rehabilitation in adults with traumatic brain injury, stroke, spinal cord injury, and musculoskeletal disorders [Granger et al., 1986; Fiedler and Granger, 1998].

Self-care items include eating, grooming, bathing, dressing the upper and lower body, and toileting hygiene. Sphincter control (continency) items include bladder and bowel management. Mobility and locomotion items include changing positions (transferring) from chairs, toilet seats and tubs, walking indoors and outdoors, self-mobility (crawl or wheelchair), and negotiating stairs. Communication items included receptive and expressive use of language, whether aural or visual (e.g., sign language, communication boards, or augmentive communication devices). Social cognitive items include peer social interaction, problem solving, and memory. The scoring system of the WeeFIM is based on a seven-level ordinal scale with high scores of 6–7 reflecting a child’s ability to complete all components of a task without adult help or supervision in a safe and timely manner. Low scores of 1 or 2 reflect that the child requires that at least half of the task components be performed by an adult. Table II lists the complete scoring codes of the WeeFIM.

In pilot studies, the WeeFIM was used to assess more than 400 children without disabilities aged 6 months to 7 years [Msall et al., 1994a]. These children were seen in primary care and community and education settings. There was a progressive increase in levels of self-care, mobility, and communication competency. Figure 1 illustrates the 10% and 90% profiles of the WeeFIM scores from the normative population of children aged 2–7 years in self-care/sphincter control (A), mobility/locomotion (B), communication/social cognition (C), and combined functions (D). In addition, over 700 children with developmental disabilities were seen in diverse settings, including early intervention community programs (United Cerebral Palsy, Association of Retarded Children, preschool programs for children with special needs), regional neonatal intensive care nursery follow-up programs, and comprehensive academic regionalized medical center programs for children with special health care needs (children with cerebral palsy, technology dependency, neurological disorders, genetic disabilities) [Msall et al., 1993, 1994b, 1997, 1998; Msall, 1996; Vohr and Msall, 1997]. These studies demonstrated that the WeeFIM had content- and criterion-related validity for children with motor, communicative, developmental, genetic and neurosensory impairments.

Between 1994 and 1996, psychometric properties of the WeeFIM were assessed in 200 children with neurodevelopmental disabilities [Ottenbacher et al., 1996, 1997]. The WeeFIM had excellent test–retest, interrater, and equivalence reliability. Equivalence reliability is the demonstration that two forms of a test yield equivalent results [Ottenbacher et al., 1996]. In order to enhance outpatient and community assessment efforts, equivalence reliability of the WeeFIM was established for one-on-one interviews and phone interviews. Criterion validity was excellent with the Vineland Adaptive Behavior Scales and the Battelle Developmental Inventory, two widely used psycho-educational assessment instruments. Last, the WeeFIM had excellent caregiver correlation with the Amount of Assistance Questionnaire.

<table>
<thead>
<tr>
<th>TABLE II. WeeFIM Scoring System</th>
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<tr>
<td>Heading</td>
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<tr>
<td>No helper</td>
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<tr>
<td>Helper</td>
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FUNCTIONAL ASSESSMENT IN GENETIC DISABILITIES

In order to illustrate the NCMRR disablement models for children with genetic developmental disabilities, functional skills and functional limitations in children with Down syndrome, spina bifida, congenital limb anomalies, congenital heart disease, urea cycle disorders, and severe developmental disabilities are described. DiGeorge malformation sequence was chosen to illustrate the spectrum of developmental phenotypes and broad categories of functional outcomes.

DOWN SYNDROME AS A MODEL OF COMMUNICATIVE-COGNITIVE DEVELOPMENTAL DISABILITY

Significant advances have occurred in prenatal risk assessment, rapid chromosome diagnostic technologies, and the molecular understanding of chromosome 21 [Holtzmann et al., 1996; Rose, 1996]. Explaining the impact of trisomy 21 on the child’s development remains the key art and science of genetic and developmental diagnosis [Cunningham et al., 1984]. A starting point for understanding developmental outcomes is to phrase functional concerns in easy-to-understand questions. These include the following:

- Will my child walk?
- Will my child talk?
- Will my child become independent in self-care, feeding, dressing, and bathing?
- Will my child be toilet trained?
- Will my child attend school with peers and learn?
- Will my child read and make developmental progress?
- Will my child have friends?
- Will my child be healthy?
- What supports will my child require to be successful? (As a parent, will I receive help if my child has developmental challenges?)

Melyn and White [1973] addressed several of these issues by using Gesell milestones in 612 children with Down syndrome living in the community. Ninety-five percent of children with Down syndrome were competent at sitting balance and self-mobility by age 2 years, 95% could walk independently by age 3.5 years, 95% were toilet trained by age 5.5 years, and 95% were speaking in phrases by age 6 years. Thus, basic mobility, continence, and communication were established before first grade. Msall and colleagues examined the use of the WeeFIM with 28 school-age children with Down syndrome (mean age, 8.8 years; range, 6–14 years) [Msall et al., 1994b]. Figure 2 illustrates WeeFIM polar graphic scores for a 6.5-year-old girl with Down syndrome. Children with Down syndrome were completely independent (median score 7) in changing positions, mobility, and bowel conti-
ence. Some supervision and assistance were required for bladder continence and dressing (for fasteners). Major functional challenges were present in expressive communication. Developmental milestones, parental expectations of educational vocational success, and intensity of special education classroom size significantly correlated with total WeeFIM score ($P < 0.05$).

An added advantage of tracking functional status is that decline in performance warrants comprehensive evaluations for biomedical conditions that interfere with developmental competencies. Thyroid disorders, symptomatic atlanto-axial instability, obstructive sleep apnea, hearing loss, visual changes, and new-onset seizures may be associated with developmental regression in children with Down syndrome [Roizen, 1996]. A further advantage of using functional assessment is that once basic functional competencies are achieved, strategies for attaining more complex educational, social, and community skills can be prioritized. Combining developmental and functional outcome studies, one could state that the average child with Down syndrome at age 2 years sits, has self-mobility, engages in gesture language, and understands some requests [Msall et al., 1991]. At kindergarten entry, the child with Down syndrome walks, talks in phrases or short sentences, is toilet trained, and is beginning to dress himself or herself [Pueschel, 1984]. In the elementary school years, the child with Down syndrome is independent in most tasks of self-care and negotiates all community motor environments, but struggles with complex language understanding and application of these skills to academic areas of reading and writing [Carr, 1995; Rondal et al., 1996].

**SPINA BIFIDA AS A MODEL OF MOBILITY AND CONTINENCY DISABILITY**

Despite advances in prenatal diagnosis and multivitamin/folate use, spina bifida remains the most common central nervous system malformation disorder [Hall and Solehdin, 1998; Olney and Mulinare, 1998]. Over the past three decades, there have been significant advances in managing hydrocephalus, neurogenic bowel and bladder, and orthopedic deformities [Sandler, 1997]. Shurtleff [1986] has provided guidelines based on the Seattle experience, summarizing mobility, self-care, and continency skills in children with spina bifida. Shurtleff demonstrated that by kindergarten entry, self-mobility, basic hand function, and complex communicative skills were present. In order to understand the impact of spinal cord dysfunction on functional skills in self-care, continency, and mobility, the WeeFIM was used in 20 children with spina bifida aged 4–10 years (Fig. 3) [Msall et al., 1994b]. A WeeFIM func-

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**A starting point for understanding developmental outcomes is to phrase functional concerns in easy to understand questions such as “Will my child walk?”**
tional quotient (FQ) was calculated using the formula:

\[
FQ = \frac{\text{Raw domain score}}{\text{Maximum domain score}} \times 100
\]

WeeFIM profiles of children aged 4–6 years were compared with WeeFIM profiles of children aged 7–10 years. Two major findings emerged. First, older children had significantly higher FQ mean scores than the younger children, and, second, children with lower-level lesions (L-3 to sacral) had significantly higher WeeFIM mean values than those with higher-level lesions. More recently, developmental functional quotients (DFQ) have been calculated using the following formula [Uniform Data System for Medical Rehabilitation, 1998]:

\[
DFQ = \frac{\text{Raw domain score}}{\text{Domain score for normal peers}} \times 100
\]

### TABLE III. Psychometric Properties of WeeFIM, Vineland, Battelle, and Amount of Assistance Questionnaire*

<table>
<thead>
<tr>
<th>Test purpose</th>
<th>WeeFIM</th>
<th>Battelle</th>
<th>VABS(^a)</th>
<th>AAQ</th>
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<tbody>
<tr>
<td><strong>Domains</strong></td>
<td>Self-care, mobility, communication—social cognition</td>
<td>Daily living (Adaptive), motor, communication, cognitive, personal-social</td>
<td>Daily living skills, motor, communication, socialization</td>
<td>Developmental skills, specialized therapies, activities of daily living</td>
</tr>
<tr>
<td><strong>Reliability</strong></td>
<td>Excellent test–retest and interrater reliability, equivalence reliability of phone interview</td>
<td>Test–retest and interrater good to excellent</td>
<td>Split half for domains and composite, excellent; test–retest, excellent; interrater, good</td>
<td>Excellent pilot reliability</td>
</tr>
<tr>
<td><strong>Validity</strong></td>
<td>Content validation by expert group; concurrent validity with VABS, AAQ, and Battelle; total WeeFIM at kindergarten entry correlates with special education resources</td>
<td>Excellent correlation with VABS; concurrent validity with WISC-R</td>
<td>Excellent correlation with school-age IQ</td>
<td>Personal assistance required by child over time directly relates to parents perception of severity of disability</td>
</tr>
<tr>
<td><strong>Application</strong></td>
<td>Measure of functional status at age 5.5 years in very-low-birth-weight–CryoROP cohort; after early intervention functional goal setting in children with cerebral palsy, spina bifida, genetic and acquired disabilities</td>
<td>Normal and disabled preschooler in early intervention; children with developmental disabilities in early elementary school</td>
<td>Preschool and long-term outcome in very-low-birth-weight children; use for all developmental disabilities</td>
<td>Children with technology dependency or major motor disabilities</td>
</tr>
</tbody>
</table>

**Administration time**
- WeeFIM: 20 min
- Battelle: 45 min
- VABS\(^a\): 30 min
- AAQ: 10 min

*From Msall [1997].

\(^a\)VABS, Vineland Adaptive Behavior Scale; AAQ, Amount of Assistance Questionnaire; WISC-R, Wechsler Intelligence Scale for Children—Revised. Battelle, Battelle Developmental Inventory; Cryo-ROP, cryosurgery for Retinopathy of Prematurity.
Figure 2. Polar graphic of WeeFIM scoring for a child with Down syndrome. Reprinted with permission of Uniform Data System & Medical Rehabilitation (UDSMR). All of the marks associated with WeeFIM belong to UDSMR.
Figure 3. Polar graphic of WeeFIM scoring for a child with spina bifida. Reprinted with permission of Uniform Data System & Medical Rehabilitation (UDSMR). All of the marks associated with WeeFIM belong to UDSMR.
The value of DFQs is that strategies to maximize current goals can be assessed from the framework of nondisabled peers. In order to understand parental priorities for independence, a mixed sample of 38 children aged 4–8 years with either diplegic cerebral palsy or lumbosacral spina bifida was assessed using the WeeFIM [Msall et al., 1992]. At a mean age of 6.4 years, 95% of the children were independent in mobility, whereas only 35% were independent in maintaining continency and 33% in self-care. Parents, however, ranked mobility as the highest current priority for their children. This study indicated that parents deferred emphasis on continency and self-care, even when their child achieved independent ambulation, because they were not confident in their abilities to help their children reach a certain level of independence. Using DFQs, strategies for encouraging appropriate current priorities can be established.

In order to promote functional skills in children with spina bifida prior to adolescence, Msall et al. [1995] performed a prospective study of 30 children with spina bifida attending a 6-week independence program for 2½ hours weekly. The curriculum included emphasis on continency, transfers, and activities of daily living as well as parent counseling strategies to facilitate age-appropriate independence of the children at home and in school. Children were administered a baseline WeeFIM and a follow-up WeeFIM 2–4 months later. Goals were determined based on WeeFIM scores and consensus of the parent, child, and therapist. Peer, same-gender teens with spina bifida were successful in teaching several younger children to be competent in intermittent catheterization. Children younger than 85 months of age changed more than children older than 85 months. Children with neurologic impairment at the T10-L2 levels had significantly lower WeeFIM scores than children with neurologic impairments at the L3 to sacral levels. The use of functional assessment allowed for a common language so that children, families, and health and rehabilitation professionals could specify priorities and attain measurable successes.

**A further advantage of using functional assessment is that once basic functional competencies are achieved, strategies for attaining more complex educational, social, and community skills can be prioritized.**

One of the advantages of functional assessment is that tasks can be completed with rehabilitation devices. Typical devices include hearing aids, mobility aids, and communicative devices. The WeeFIM was used to assess 32 children aged 4–11 years who required specialized prostheses because of limb deficiencies [Msall et al., 1994b]. Ninety percent of the children needed prostheses had congenital limb-reduction deficiencies. The mean total WeeFIM DFQ was 88%, reflecting competency levels in self-care, mobility, and communication/social cognition. All children with lower-limb deficiency walked with prostheses. Twenty-two percent of the children required some assistance in self-care activities, reflecting the complexity of bimanual skills required in manipulating zippers, snaps, and buttons during dressing tasks. Ninety percent of children were in regular educational facilities. Children who fed themselves early on had significantly higher total WeeFIM scores than children who learned to self-feed later. Thus, initial developmental self-care skills were associated with subsequent total WeeFIM scores in disorders where the prognosis in mobility and communication developmental skills is good. The importance of functional assessment in children with limb anomalies is that successful use of prosthesis results in mobility independence for children with lower-limb deficiencies, self-care independence for children with upper-limb deficiencies, and regular education successes as the result of communicative and social cognitive competency.

**HYPOPLASTIC LEFT HEART AND SINGLE-VENTRICLE AND CONGENITAL HEART DISEASE**

Major advances in prenatal and postnatal diagnosis and cardiac surgery have led to wider availability of interventions for children with congenital heart disease. Certain life-threatening cardiac malformations can be viewed as natural experiments for assessing reconstructive approaches. Rogers et al. [1995a,b] studied the neurodevelopmental and functional status of children who underwent surgical interventions for hypoplastic left-heart syndrome (HLHS) or single-ventricle heart anomaly (SVHA). None of the survivors had a known chromosomal or dysmorphic syndrome. Eleven infants with HLHS (mean age at follow-up, 3.1 years) and 15 infants with SVHA (mean age at follow-up, 5.5 years) were given psychometric [Bayley, McCarthy, Weschler Intelligence Scale for Children—3], neurodevelopmental, functional (WeeFIM), and physical examinations. Cerebral palsy was present in 18% of children with HLHS and in 0% of those with SVHA. Mental retardation was present in 73% of children with HLHS and in 0% of those with SVHA. Results showed that 20% of children with SVHA had functional limitations, whereas 73% of the HLHS children were functionally impaired. Of the seven children with SVHA assessed between ages 3 and 6 years (mean age of follow-up, 3.9 ± 1 years), the mean total WeeFIM score was 86.6 ± 9.5, an age equivalent of 37 months [Uniform Data System for Medical Rehabilitation, 1998]. Of the seven HLHS survivors aged 3–6 years (mean age of follow-up, 4.2 ± 1.1 years), the mean total WeeFIM score was 45.4 ± 24, an age equivalent of 19 months [Uniform Data System for Medical Rehabilitation,
Figure 4. Polar graphic of WeeFIM scoring for a child with hypoplastic left-heart syndrome. Reprinted with permission of Uniform Data System & Medical Rehabilitation (UDSMR). All of the marks associated with WeeFIM belong to UDSMR.
1998). Thus, by analyzing functional outcomes, one is able to examine physiological factors that preserve central nervous system integrity in SVHA patients compared with factors that impair central nervous system functions responsible for age-appropriate functional skills in children with HLHS. Figure 4 is a WeeFIM polar graph of a 4.5-year-old child with HLHS, and demonstrates significant self-care and cognitive functional limitations.

NEURODEVELOPMENTAL AND FUNCTIONAL OUTCOME IN UREA CYCLE DISORDERS

Children with congenital urea cycle disorders who survived neonatal hyperammonemic coma were assessed in early childhood [Mall et al., 1984]. Children with more prolonged hyperammonemic encephalopathy, especially if longer than 72 hours, had lower developmental scores and more central nervous system abnormalities on CT neuroimaging. Seventy-nine percent of children had mental retardation, 46% had cerebral palsy, 4% were blind, 17% had epilepsy, and 46% had multiple neurodevelopmental impairments.

Fourteen children with late-onset (partial) urea cycle disorders (partial enzyme activity) were also examined [Mall et al., 1988]. The mean number of hyperammonemic episodes was four (range, 0–11). Rates of neurodevelopmental impairment were high: 57% had mental retardation (mean IQ, 56; range, 10–103), 33% had cerebral palsy, 20% had epilepsy, 13% had cortical blindness, and 33% had multiple neurodevelopmental impairments. In children with prolonged neonatal hyperammonemic coma and more severe late-onset hyperammonemic coma, motor, communicative, and self-care limitations were high. Both findings suggest that accurate and early diagnosis is essential in minimizing the preschool morbidity of substantial functional limitations in children with urea cycle disorders.

Maestri et al. [1999] have recently reported developmental outcomes of a cohort of boys with neonatal onset of ornithine transcarbamylase (OTC) deficiency, seen between 1976 and 1996 at Johns Hopkins University Hospital. Neonatal mortality was 46%. Among the 28 children whose developmental outcomes were known, 16 had severe to profound neurodevelopmental impairments and subsequently died. Three children received liver transplants, two of whom had major neurodevelopmental disabilities before transplant. Of the nine long-term survivors with developmental data, five had severe mental retardation with developmental quotients (QD) < 50, and three had mild to moderate mental retardation (DQs of 50–70). In addition, five needed tube feedings, four had seizure disorders, three had spastic quadriplegia, and two had microcephaly. Thus, even with current efforts of metabolic rescue and maintenance, high mortality and severe motor, self-care, and communicative limitations are present in OTC neonatal survivors.

CHILDREN WITH SEVERE DEVELOPMENTAL DISABILITIES: IMPACT OF BASIC FUNCTIONAL SKILLS ON MORTALITY

Eyman and colleagues [1990] examined a cohort of severely developmentally disabled children. One example of a known severe impairment in this cohort would be trisomy 18. In initial studies, they conclude that approximately 20% had genetic origins. As mobility, feeding skills, and hand skills increased, mortality decreased. Strauss et al. [1997] reexamined the original data and included children who showed improvements in early functional skills. He separated the children into four groups. Group A children were tube fed and unable to lift their heads; Group B were tube fed and able to lift their heads, with limited rolling; Group C were not tube fed and unable to lift their heads; and Group D were tube fed and capable of rolling/sitting or were not tube fed and able to lift their heads. The five-year survival rates were 55% for Group A, 69% for Group B; 69% for Group C, and 94% for Group D. The ability to perform some motor functions, with postural change, hand skills, or feeding skills was a huge predictor of mortality in the severely developmentally disabled child. Additionally, the inability to chew, suck, and coordinate swallowing may be an indicator of brain-stem dysfunction with significant impact on long-term outcome. These studies demonstrated that severe functional limitations contribute to both quality and quantity of life in genetic, perinatal, postnatal, and unknown impairments.

DIGeorge MALFORMATION SEQUENCE AS A MODEL HEARING AND DEVELOPMENTAL DISORDER WITH CRANOFACIAL AND CARDIAC MALFORMATIONS

Advances in molecular genetics have allowed for a better understanding of contiguous gene chromosomal disorders. Chromosome 22 was found to be a key region associated with craniofacial and cardiac malformation and is illustrated by the DiGeorge Malformation Sequence. Through the initial neonatal priorities, including managing complex cardiac, endocrine, and immune system dysfunction, audiological and neurodevelopmental assessment are key characteristics affecting long-term outcome. The spectrum of DiGeorge and velocardiofacial malformations includes strengths in motor and self-care skills but challenges in expressive speech and language-related learning disabilities [Driscoll et al., 1996]. However, if there is significant sensorineural hearing loss, then the habilitative management goals are for children with deafness. Advances in neonatal audiological screening using transient oto-acoustic emissions and automated brainstem responses have led to earlier recognition of sensorineural hearing impairments in all children [Vohr et al., 1998]. Earlier identification of children with significant sensorineural hearing impairments will allow for appropriate family supports and educational strategies [Windmill, 1998]. Accardo [1999] has recently emphasized the importance of the spectrum of developmental disabilities in a variety of genetic disorders. Accardo and Capute [1998] have high-
CONCLUSION
Appropriate use of functional assessment tools will allow clinicians to describe children’s strengths and challenges across health, developmental, educational, and community settings.

ACKNOWLEDGMENTS
The authors gratefully acknowledge the efforts of Brian T. Rogers, Thomas M. Lock, Nancy Lyon, Sue LaForest, Ken Ottenbacher, Germaine Buck, and Kathleen DiGaudio at the Robert Warner Rehabilitation Center for Functional Assessment, SUNY at Buffalo, for their dedicated efforts in measuring functional challenges in children with developmental disabilities. This article is dedicated to Dr. Arnold Capute in celebration of his lifelong efforts to promote comprehensive neurodevelopmental and functional assessment systems for children with genetic and developmental disabilities. This paper was supported in part by MCH Behavioral Training Grant MCJ 449505-02-0.

<table>
<thead>
<tr>
<th>TABLE IV. Recognizing Developmental Phenotypes—Levels of Functional Disability</th>
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<tr>
<td><strong>Level of disability</strong></td>
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<tr>
<td>Mild Developmental Disability: WeeFIM scores within 1.5 SD of the mean for age group</td>
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<tr>
<td>Moderate developmental disability: WeeFIM scores 1.5–2.5 SD below the mean for age group</td>
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<tr>
<td>Severe developmental disability: WeeFIM scores 2.5–3.5 SD below the mean for age group</td>
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<tr>
<td>Profound developmental disability: WeeFIM scores 3.5 SD below the mean for age group</td>
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lighted the complexity of syndrome impact on developmental diagnosis. Overall, recognizing developmental phenotypes and their functional impact is summarized in Table IV.

can focus on factors that promote literacy, friendships, handling of information, and community participation. Important outcome areas in middle childhood include grade repetition, special educational supports, and successes outside school. Vital areas of community participation include sports, scouts, music, church, clubs, and hobbies. In children with major challenges in mobility and self-care, supports to families for respite, visiting nursing, and personal care assistance; supplemental security income; Early Periodic Screening, Diagnosis, and Treatment (EPSDT); and the Katy Beckett Community Waiver Program can be highlighted. If a child is significantly challenged, the importance of comprehensive family supports and access to quality protocols for positioning, feeding, and self-care are necessary. In children who have motor abilities but challenges in communication and social cognition, community and family supports can be prioritized for respite, home services, and community participation. In this way, the complexity of genetic disorders on the central nervous system can allow for a perspective of the child’s strengths and challenges and family strengths and supports. By describing the complexity of genetic mechanisms on developmental and functional status, medical professionals can optimize quality of life, provide family support, and critically contribute knowledge to preventive efforts.
REFERENCES


