

# Caregiver assistance in treating mitochondrial diseases in children

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## OBJECTIVE:

The aim of this study was to evaluate the caregiver assistance in a cohort of children with mitochondrial diseases.

## METHODS:

Data of 22 children with mitochondrial diseases, who showed significantly decreased activity of respiratory chain enzymes and/or pyruvate dehydrogenase complex in a muscle tissue, was evaluated (Table 1). All children were scored by Mitochondrial Disease Criteria as a »definite« mitochondrial disorder (1). Follow-up period was from 15 months to 18 years. The Pediatric Evaluation of Disability Inventory (PEDI) was administered and scaled scores determined (2,3). Summary scores of the PEDI are based on an estimate of the placement of an individual child along the continuum of hierarchically sequenced item within each domain. The scaled scores (0 to 100) are reflecting an increasing level of functioning and provide an estimate of the child's achievements of activities and amount of assistance regardless of age.

Laboratory measurements of enzyme's activities were performed in the Laboratory for molecular neurogenetics Istituto Nazionale Neurologico "Carlo Besta", Milan, Italy.

## RESULTS:

The variability in caregiver assistance was large, ranging from independent functioning to totally dependent on help in Self-care, Mobility and Social Function (Figure 1, Table 2 and 3). In Self-care domain, the children were relatively better in eating (median 0,5, range 0-5), but in all other activities they were mostly completely dependent (medians 0, range 0-5). In Mobility domain, many children were mostly dependent on caregivers. Some independence was seen in indoor and outdoor locomotion (medians 1,5, range 0-5). In Social function domain children were relatively better in functional comprehension and expression (medians 2, range 0-5) then in joint-problem solving, peer play and safety (medians 1, range 0-5).

Table 3. Descriptive Statistics for Caregiver Assistance of Children with Mitochondrial Diseases

Number	Caregiver Assistance								
	Self-care			Mobility			Social Function		
	Mean	SD	Range	Mean	SD	Range	Mean	SD	Range
18	29,0	2,0	0-100	36,6	3,2	0-100	42,9	4	0-100

Table 1. Characteristics of children with mitochondrial diseases (n=22)

Age, years	
Range	0,5-18,5
Mean (SD)	7,3 (5,1)
Sex, number (%)	
Male	9 (41)
Female	13 (59)
Enzyme defect, number (%)	
PDHc	12 (55)
RC	10 (45)
Age at presentation, number (%)	
Neonatal	8 (36)
Infant	14 (64)
Cognitive function, number (%)	
Global developmental delay	8 (36)
Moderate developmental delay	4 (18)
Mild developmental delay	4 (18)
Normal cognitive function	2 (9)
Neurological examination, number (%)	
Increased muscle tonus	7 (32)
Spastic tetraparesis	2 (9)
Spastic tetraparesis with rigidity	4 (18)
Spastic diplegia	1 (5)
Decreased muscle tonus	11 (50)
Leigh syndrome	6 (27)
Molecular analysis	
Thymidine kinase 2 gene (TK2)	2 (9)
Cytochrome c oxidase (COX) assembly gene (SCO2)	1 (5)
Plasma lactate level, number (%)	
Normal	7 (32)
Increased	15 (68)
Number of deaths (%)	4 (18)

Table 2. Results of Caregiver Assistance points per question in each domain.

SELF-CARE	Median	Range
eating	0,5	0-5
grooming	0	0-5
bathing	0	0-5
dressing upper body	0	0-5
dressing lower body	0	0-5
toileting	0	0-5
bladder management	0	0-5
bowel management	0	0-5
MOBILITY		
chair/toilet transfers	0,5	0-5
car transfers	0	0-5
bed mobility/transfers	0	0-5
tub transfers	0	0-5
indoor locomotion	1,5	0-5
outdoor locomotion	1,5	0-5
stairs	0,5	0-5
SOCIAL FUNCTION		
functional comprehension	2	0-5
functional expression	2	0-5
joint problem-solving	1	0-5
peer play	1	0-5
safety	1	0-5

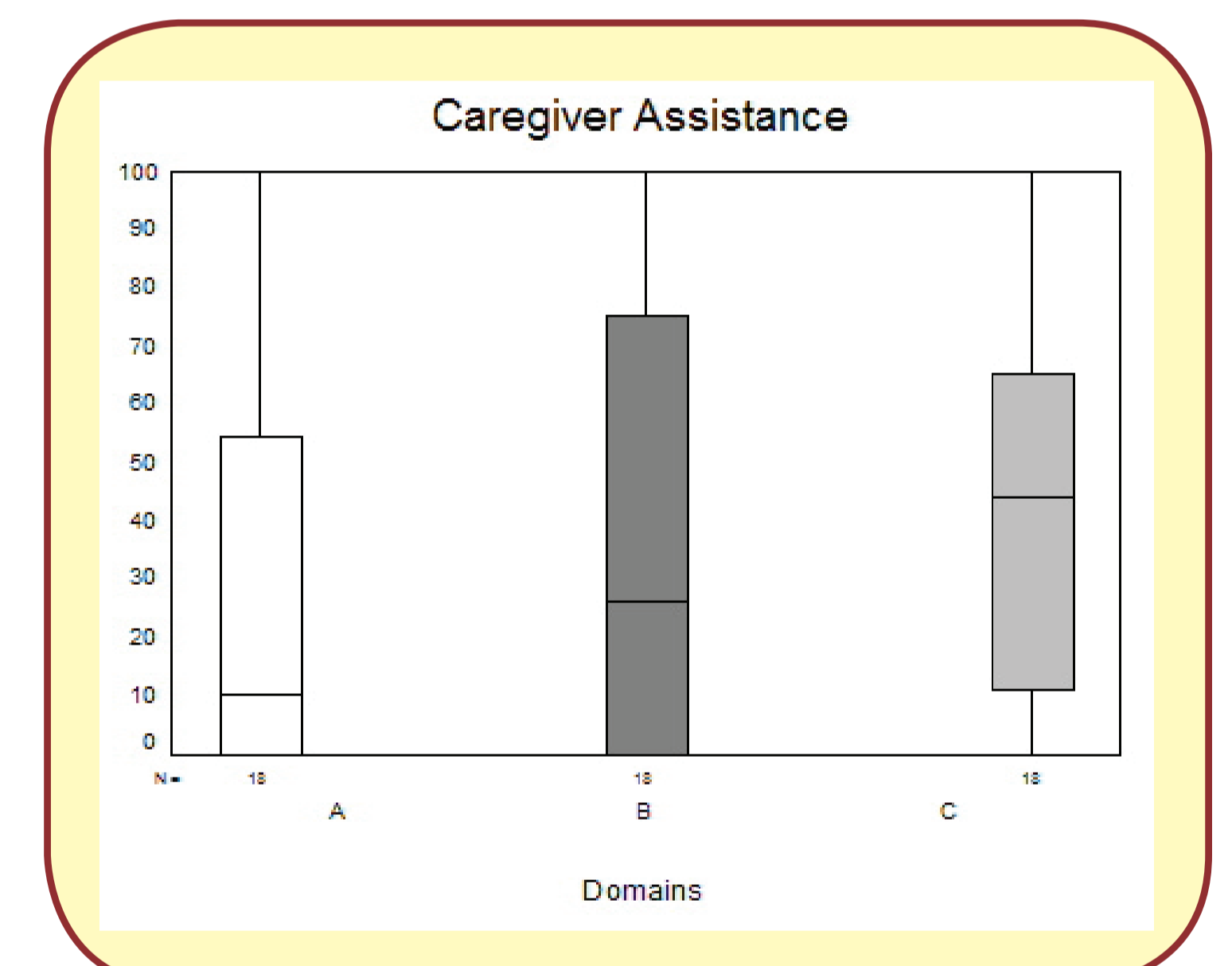


Figure 1: Box plot of Caregiver Assistance Scaled Scores (0-100) in use per child in Self-care (A), Mobility (B) and Social Function (C). Boxes cover interquartile range, squares indicate median, whiskers extend to 10th and 90th centiles.

## CONCLUSION:

Our study revealed the great importance of caregivers and/or parents in a life of a child with a mitochondrial disease. All children were dependent on parents. Stress in parents of handicapped children is especially related to child's maladaptive behaviour and parental cognitions (4,5). To treat children with mitochondrial diseases is not only to provide successful medication, but also to make a better environment for handicapped children, better rehabilitation, social conditions and also helping their parents.

## LITERATURE:

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