



## Barth Syndrome Foundation

[www.barthsyndrome.org](http://www.barthsyndrome.org)

## Transitions

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### BACKGROUND

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Since the first article reporting Barth syndrome about two decades ago,<sup>1</sup> the survival rates associated with children who Barth syndrome have improved greatly such that we are now seeing children with this disorder survive to adulthood. In general it is estimated that 1 in 10 or more individuals under the age of 17 have a chronic illness or disability.<sup>2</sup>

It is thought that an early diagnosis leading to an improved understanding about Barth syndrome, cooperation between attending physicians and experts in the area of Barth syndrome has resulted in successful therapeutic interventions reducing the mortality rate by as much as 50%.<sup>3</sup> Societal changes now encourage more adults with physical challenges to live at home with appropriate support.<sup>4</sup>

There is no longer a reason for a parent of a child with Barth syndrome to expect anything less than for their child to live a full, productive life. Pediatricians are preparing at an increased rate to play a critical role in reinforcing positive attitudes and beliefs while helping adolescents and their families to make appropriate transitions to adult health care.<sup>5</sup> It is likely that our young adults will require ongoing medical care for Barth syndrome.

The pediatrician as well as other pediatric specialists can and should play a central role in helping adolescents and our BSF families make the transition to adult health care. Toward this end, the pediatrician needs to know which practitioners in the community have the clinical skills needed and an interest in working with adults who have a complex disorder such as Barth syndrome.

Under some circumstances, young adults with special health care needs have been reported to continue with their current health care supervision because some pediatricians are comfortable with adult health. Criteria and timing for eventual transitions will vary. It is recommended to approach pediatricians about their thoughts on the issue of transferring care from pediatric to adult care. It may be prudent to obtain this information as early as when interviewing a pediatrician during infancy. Learn how flexible they are when deciding on when to transfer adolescent patients to adult facilities and ask if certain considerations are made when making this decision.

## **ISSUES IN HEALTH CARE TRANSITIONS**

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Because of the complexity of Barth syndrome it is likely that our young men will require coordinated care involving multiple health care providers/specialists. Coordination and access may be more difficult when adolescents reach adulthood; often, with other disorders an entirely new group of health care providers assume responsibility for adult patients. It is unlikely these new providers of care will be familiar with Barth syndrome, patient histories and the priorities and concerns of these young men and their families.<sup>6</sup>

Also, adolescents with Barth syndrome may need to adjust to the possible loss of a close and longstanding relationship with their pediatrician and other specialists when transferring to adult health care. This change may be difficult, because many pediatricians often work with multidisciplinary teams while adult-oriented health care providers tend to focus more on specific system-oriented complaints and rely on independent consultants for further specialty care. Because many adult health care providers receive only limited training regarding adolescent or young adult disorders associated with disabilities and transitional issues, the provider may not be familiar with Barth syndrome and its management.

Schidlow and Fiel have summarized other major obstacles to smooth transitions from adolescent to adult health care.<sup>7</sup> The severity of the illness or disability, the level of maturity, acceptance and understanding of the patient, additional environmental or family stresses, the need for control by parents or health care provider, a distorted perception (by parents or health care provider) of potential patient outcomes, and lack of patient or family support systems all may contribute to transition stress.

### **Independence and Dependence**

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The goal for all children is to move progressively from complete dependence toward independence. Children with Barth syndrome, should be encouraged to develop the highest possible level of independence based on a realistic and objective evaluation of their individual abilities and limitations. Even though a child may have Barth syndrome there are still varied degree of limitations reported.<sup>8</sup> The adolescent with Barth syndrome and the family may be unsure of the benefits of greater independence or even unaware of the patient's ability to achieve partial or total independence.<sup>9</sup> The adolescent with Barth syndrome should be involved in the process of making decisions about when and to whom they will be referred.

The transition of preparation of separation of a parent from a teenager with Barth syndrome is a difficult but essential part of long-term management and allows parents to think about their children surviving into and past adolescence. A number of factors make separation and independence more difficult for adolescents with Barth syndrome and for their parents. These include the need for treatment, parental overprotection, an appearance that is more youthful than the chronological age and limited physical freedom.<sup>10</sup>

Measures should be taken to encourage the adolescent in the involvement of care and understanding of Barth syndrome. Adolescents need to be aware of their treatment choices and be encouraged to discuss and participate in making decisions about these choices. Allowing the adolescent to take control in simple ways, such as choosing the form of medication (pill, liquid, etc.), will foster autonomy as well as improve compliance. The next step may be supervised self-administration of medications on a daily basis beginning with laying the meds out in a pill-box for daily administration, eventually leading to the monitoring the adolescent laying his own meds out in his pill-box. A useful tool to assist the Barth adolescent in the manner of taking meds on a timely basis is a multi-alarm wristwatch.

**Educating adolescents about Barth syndrome has beneficial effects:**

- ❖ They learn how to avoid situations that exacerbate their condition; such as over-stimulation
- ❖ They learn how to minimize the severity of an exacerbation; properly educated about Barth syndrome the adolescent will be less likely to do something that will cause days of fatigue
- ❖ They learn self-care skills to minimize daily effects of their illness

Learning self-care skills is an important way to enhance self-esteem and autonomy. If the adolescent can participate in his own care, self-esteem will be enhanced and the mechanics of separating eased.

For adolescents who are more dependent on others for a variety of care needs, the acquisition of some of the skills needed for self-care and independent living can contribute to a more successful and comfortable life at home. This learning promotes self-esteem and facilitates more participation in their community. Toward this end, the person with Barth syndrome should have the opportunity to acquire skills to understand and to develop a sense of financial responsibility when appropriate. The development of skills for independent living (such as living alone, planning or preparing meals, driving, recognizing the need for adequate rest or grocery shopping) requires assistance from the parents.

## **Physical Appearance**

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All adolescents are self-conscious about their physical appearance and pubertal development. Concerns about delayed growth or pubescent changes are difficult for an adolescent. Adolescents with Barth syndrome need to be involved in discussion about Tanner stage development or results of bone age measurements with health-care providers. It may be beneficial to consider a peer-to-peer discussion between the adolescent and a young adult with Barth syndrome who has passed through this difficult period. This has been reported to be an emotionally difficult time for the adolescent Barth male. He is seeing his healthy peers involved in sports, developing and reaching stages of development earlier. Healthy dialog between a trusted psycho-social therapist, educator, minister or physician may also be found beneficial. Anyone who the young

teen admires and finds as a source of encouragement may be found to be a valuable resource during this difficult time.

## **School and Work**

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Healthy teenagers spend a large part of their day in school and later at work. Attention should be paid to the concerns of the adolescent with Barth syndrome about absenteeism resulting from appointments and complications of the various components of the disorder. Meetings with teachers should be made before the school year begins; providing literature about Barth syndrome, direction to the BSF website and encouragement to interact with other educators who are experienced in working with those who have Barth syndrome.

Some adolescents are reported to have extreme fatigue during these years often requiring a special approach to education. Parents need to be realistic in their expectations for adolescents with Barth syndrome. It is important not to expect the achievement of the adolescent with Barth syndrome to be beyond the teen's abilities. It is equally important not to settle for goals much below the teenager's potential.

Most adolescents with disabilities other than Barth syndrome have participated in modified educational programs. Frequent and prolonged hospitalizations or illness convalescence have been reported to interrupt continuity or delay the achievement of educational goals. Many well-intentioned programs have been reported to offer limited opportunities for experiences outside the school and may fail to prepare the adolescent for many life experiences and the world of work and independent living.<sup>11</sup>

Preparation for vocational assessment and training should not be postponed until the student is 18 to 21 years old because training may be prolonged for the adolescent with a Barth syndrome. It is important for the family, and high school advisor to seek assistance for the adolescent, including but not limited to the understanding of the medical aspects of the student's condition as well as seeking an appropriate person to coordinate the evaluation and transition to higher education or vocational training.<sup>121314</sup>

In the US there is a nationwide federal/state vocational rehabilitation program, which has an agency in each state capital as well as other local offices. These locations can be found in the telephone directory (under the state listing for Rehabilitative Services or Vocational Rehabilitation Services.) These agencies assist eligible people with disabilities to define a suitable employment goal, assist with additional educational opportunities, and assist with employment. Supported or sheltered employment options provide opportunities that can enhance the quality of life and personal satisfaction of young people with disorders such as Barth syndrome.<sup>15</sup> The Americans With Disabilities Act also has the potential to help with issues of access to job opportunities and public facilities.<sup>16</sup>

Assurance that a goal can be reached is important with the adolescent or young adult with Barth syndrome. Delays are okay. It is understandably frustrating for those who have

Barth syndrome to see his peers ahead of him in school or obtain gainful employment before him. Positive approach to education and careful realistic consideration to vocation can assist in obtaining reachable goals one-step at a time. While it may be a challenge it is possible for adolescents and young adults with chronic illnesses to find meaningful employment.<sup>17</sup>

## **Insurance Issues and Limitations**

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Adolescents with chronic illnesses such as Barth syndrome face special problems regarding access to health insurance, especially when they are no longer covered by their parents' insurance.<sup>18 19</sup> Currently, insurance provisions regarding preexisting medical conditions may limit or even preclude insurance eligibility for those who have Barth syndrome in the US. It is important to begin seeking out insurance coverage for the transition long before the time the adolescent is no longer covered under the parent's policy. Review the insurance policy, call the carrier to find out what language would be required to maintain insurance on the individual with Barth syndrome. Don't wait until the Barth adolescent is 17 to inquire about this. Early intervention and appeals may make a difference in obtaining a positive change for your child's insurance benefits. Advocating for policy changes takes time, patience and perseverance. Act early and be on guard for any policy changes that could exclude benefits for your child.

The adolescent or young adult with disabilities often requires insurance that covers a broad range of services, including multiple medical specialty consultations, laboratory tests, equipment, and prescription plans.<sup>20</sup> Such insurance rarely is available to many workers with disabilities, which may limit their productivity on the job or the types of jobs for which they are qualified, forcing them to accept lower-wage employment that may not offer insurance or provide the types of benefits needed at an affordable cost.

## **Social Security**

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In the US The Social Security Administration directs two programs of financial assistance to eligible persons during the transition process.<sup>21 22</sup>

First, the Supplemental Security Income program provides cash benefits for low-income persons who are blind or have other disabilities. Recent Supplemental Security Income changes help children and those younger than 18 years with disabilities to qualify for benefits. Persons older than 18 years are eligible to receive monthly payments if they have little or no income or resources (eg, a savings account) and have a substantial disability.

Second, the Social Security Disability Income considers the employment status of the applicant's parents. "Benefits are paid to persons who become disabled before age 22 if at least one of their parents had worked a certain amount of time under Social Security but now is disabled, retired, and/or deceased."

Recent legislation has made major changes in both programs to encourage people with

disabilities receiving these benefits to work and to become independent. Also, certain provisions of Supplemental Security Income allow for the maintenance of Medicaid coverage even when the young adult becomes employed.<sup>23</sup>

Information is available through the local Social Security office or by calling the Social Security Administration toll free at 1-800-772-1213 (voice) or 1-800-325-0778 (TDD).

## **Hospitalization**

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Young adults with Barth syndrome may require frequent hospitalization, just as they did during childhood. As they mature, many develop ambivalent feelings regarding hospitalization, particularly when they become the oldest patients in the pediatric or adolescent unit. At the same time, however, their familiarity and comfort with the unit personnel, policies, and procedures may make them reluctant to enter an adult unit. Inpatient care should be a part of the short- and long-term planning for the management of the adolescent and eventual young adult with Barth syndrome. Hospitalization may be an essential or infrequent adjunct to a health care program but still is subject to issues of transition and planning. Preferably, arrangements for hospital care should not be made on an urgent or emergent basis but planned well in advance with attention to detail.

The individual plan for hospitalization should consider whether the adolescent or young adult prefers admission to an adolescent or adult unit. It is important that the unit staff receive training in the management of Barth syndrome. When pediatricians continue to provide primary care for young adult Barth patients, they should be requested to seek admitting privileges to the adult unit to ensure their continuing participation as the primary attending physician or as a consultant.

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## **SUMMARY AND RECOMMENDATIONS**

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Because of a better understanding about Barth syndrome and improved care for children diagnosed with Barth syndrome more children with this disorder are now reaching adulthood. Planning is essential to achieve appropriate transition from pediatric health care to adult health care.

### **Transition planning should:**

- ❖ Begin early, with special attention to maximizing opportunities for independence and for the necessary health, educational, and social services;
- ❖ Include active participation of the family, healthcare team and Barth adolescent in the process;
- ❖ Consider the Barth adolescent individually, realistically, and positively, encouraging functional independence and appropriate attitudes toward self-worth and interpersonal relationships (including issues of sexuality);
- ❖ Encourage the Barth adolescent's willingness or ability to accept the plan;
- ❖ Consider having the pediatrician and the adult health care practitioner as co-managers for a period of time (eg, 1 or 2 years); and include recommendations by

- the pediatrician for referral to adult health care providers (especially sub-specialists) who are sensitive to and have an interest in families that include adults with special health care needs.
- ❖ The healthcare team and Barth family should participate actively in the above process and become aware of local, state, and national resources for assistance to ease the transition of the Barth adolescent
  - ❖ Above all, the pediatric health-care providers and families should work closely as advocates in the transition of childhood to issues surrounding chronic illness in an adult.
  - ❖ Healthcare providers, educators and resource personal used throughout childhood should be enlisted for assistance to adapt positively to an adult-focused system of health care.

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