OVERALL CONCLUSIONS

Nature of Heritable Disorders of Connective Tissue

1. Heritable disorders of connective tissue (HDCTs) comprise a large and varied group of disorders in children and adults that share the common feature of pronounced involvement of connective tissues, usually in multiple organ systems. HDCTs can lead to a variety of physical and mental secondary impairments (i.e., manifestations, medical diagnoses, syndromes, comorbidities, or other health conditions) and associated functional limitations. Impairments can range from minor to severe and even life-threatening and may fluctuate in severity over time in an individual. Functional limitations may be sufficiently severe to interfere with participation in work and school, as well as social and recreational activities, and may include precautions and restrictions on activities to avoid aggravating the condition.

Heritable Disorders of Connective Tissue and Disability

2. Some of the Social Security Administration's (SSA's) Adult Listings apply directly to secondary impairments experienced by individuals with HDCTs and could be used to evaluate disability in those individuals. Other listings, with some modification, could apply to certain secondary impairments experienced by individuals with HDCTs.

Diagnosis

3. Early diagnosis of HDCTs is important to reduce physical injury, reduce psychological harm to the individual and family members, and prevent the risks associated with inappropriate or fragmented medical care.
   - Diagnosis of HDCTs is often delayed because of
     - the multisystem, complex, and phenotypically variable nature of the disorders;
     - lack of knowledge about HDCTs among health care providers, patients and family members, and other stakeholders;
     - lack of experience with using the syndromic approach to diagnosis, in which diagnosis is based on characteristic groups of symptoms and signs;
     - lack of access to comprehensive, multidisciplinary care teams with expertise in HDCTs (due to a shortage of clinicians, especially in some geographic areas);
     - historical bias and denial among health care providers, patients, and family members about the reality of the lived experience of manifestations of the disorders; and
     - inaccurate expectations that there will be a diagnostic genetic test for every HDCT.
   - Delayed or misdiagnosis of individuals with HDCTs can result in
     - inappropriate medical interventions;
     - inability to accurately assess the risks and benefits associated with medical procedures;
     - inability to access necessary reasonable accommodations at work or school;
     - family stress and dysfunction;
     - stress associated with unexplained and repeated evidence of trauma, leading to inappropriate suspicion of child abuse;
     - inappropriate assessments and incorrect diagnoses; and
     - mistrust of health care providers and negative expectations for future health care encounters.
   - Timely diagnosis and recognition of the many physical and mental secondary impairments with which HDCTs can present and action to address them, even if in the absence of a specific molecular diagnosis, can dramatically improve individuals' quality of life and functional status, including the ability to participate in work and school.

Management

4. Although curative treatments for HDCTs do not exist at this time, appropriate understanding and management of the disorders can reduce the frequency and severity of their manifestations and resulting functional consequences. High-quality care for individuals with HDCTs relies on effective coordination among a team of clinicians across a broad range of physical and mental health care disciplines who are knowledgeable about these disorders.

Barriers to Access to Care

5. Access to comprehensive, multidisciplinary care for the diagnosis and management of HDCTs can be limited by geography and other factors, including the availability of care teams with expertise in the disorders.

Education

6. Education about HDCTs, including their multisystem manifestations, diagnosis, and management, is important for all clinicians to help increase recognition and earlier diagnosis of the disorders and enable the provision of appropriate care.
   - A variety of health care providers should be able to recognize HDCTs and direct affected individuals to the appropriate clinicians for management.
   - Individuals with HDCTs and relevant support groups can provide valuable insight regarding the manifestations and lived experience of the disorders.
   - Increased recognition of the breadth and scope of HDCTs by health care professional education programs, professional organizations, and publishers of quality biomedical research is needed.

7. Education of individuals with the disorders and their families, as well as employers and school staff, is important to improve the quality of life for affected individuals and their families, to facilitate appropriate accommodations at work and school, and to help inform the disability assessment and determination process.

Research Gaps

8. Ongoing research on HDCTs is important to advance understanding of the disorders and their effects. In particular, research on care services and interventions for HDCTs and secondary impairments is needed, including
   - more specific diagnostic criteria and biomarkers;
   - functional and biomeasures of severity;
   - effective treatment for HDCTs and management of their physical and mental manifestations, including comparative treatment trials;
   - the clinical course of the disorders throughout the lifetime of affected individuals;
   - the impact of relevant reasonable accommodations on affected individuals' ability to participate in work and school; and
   - benefits versus risks of participation in common childhood activities (e.g., contact sports, gymnastics, dance).

To read the full report, please visit
http://www.nationalacademies.org/connective-tissue-disorders

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