



## CARING FOR THE SUDC FAMILY: MEDICAL AND BEREAVEMENT INFORMATION FOR THE CLINICIAN'S CONSIDERATION

When a child dies unexpectedly, a family may feel a flood of emotions, including guilt, anger and lack of control. The clinician's role is to offer stable support, information and compassionate care. In order to provide the most appropriate care for families in these situations, it is necessary to understand the complexities of the grief and trauma response and to recognize that the families' needs will change over time.

The following suggestions include considerations for both the medical and psychosocial needs of the primary family members. These provide some general guidelines and recommendations to consider, recognizing that they will need to be tailored for each individual family based on the specific circumstances of their child's death, significant findings from the death investigation, the child/family medical history, the clinical evaluations of family members and their family circumstances.

*Disclaimer: This information is not meant to diagnose or treat. The information contained is general and therefore not specific nor comprehensive to meet the needs of any given family. It should not be construed as medical advice but supportive information to assist the primary care physician. Please ask your physician if you have medical questions pertaining to your, or your child's health.*

1. Introduction
2. Recommendations for Communicating with Families
3. Meeting the Mental Health Needs of Family Members Affected by Sudden Death
  - a. Emotional and Psychological Symptoms of Trauma
  - b. Therapeutic Interventions
  - c. Recommendations on Supporting Siblings
  - d. When to Refer for Professional Support
  - e. Resources
4. Medical Care Considerations of Family Members Following Sudden Death
  - a. Recommended Follow up from Primary Care Provider
  - b. Considerations for Acute Care of SUDC Siblings
  - c. DNA Banking
  - d. Evaluating for Inborn Errors of Metabolism
  - e. Evaluating for Cardiac Channelopathies
  - f. Evaluating for Seizures
5. Note to Parents and how to use this document

Last Modified April 2019



## 1. Introduction

Grief is the natural reaction to loss. Grief is both a universal and a personal experience. It is unique to the individual. Most people will experience this natural process and not require professional assistance to integrate it into their lives. Having a strong support network is associated with positive outcomes.

Trauma is defined by the Center for Anxiety and Mood Disorders as the “psychological, emotional response to an event or an experience that is deeply distressing or disturbing.” Reactions to trauma are also individualized, but commonly include intrusive re-experiencing of the event, avoidance of trauma-related reminders, hyperarousal, and strong negative emotions such as fear, anger, guilt, and shame. These reactions are common after a traumatic event and typically diminish over time. Psychologists categorize persistent trauma reactions as post-traumatic stress disorder when symptoms are ongoing past the first 1-3 months.

When a child dies unexpectedly, no one is prepared. Children are supposed to outlive their parents. A child’s death defies the natural order of life. It is normal for family members to experience both intense grief AND trauma. Clinicians have an important role, to provide ongoing follow-up of families, monitoring their needs over time and recognizing warning signs that may require professional intervention.

For more information on SUDC Facts:

The SUDC Foundation website at: [www.sudc.org](http://www.sudc.org)

The National Association of Rare Disorders: <https://rarediseases.org/rare-diseases/sudden-unexplained-death-in-childhood/>

## 2. Recommendations for Communicating with Families

- Although there are some gender stereotypes and common cultural traditions in coping with death, remember that no two individuals grieve alike.
- During an open investigation, do not speculate on the cause of death with the family. Although this may feel supportive, it often undermines the relationship with the Medical Examiner/Coroner who is legally charged with determining cause of death and adds stress for the family when clinicians and the investigating authorities disagree. It is optimal that clinicians and Medical Examiners/Coroners communicate directly for the best interests of the family.
- Grief is exhausting. Recognize that the capacity to participate in medical discussions may be limited. Provide instructions verbally and in writing. The ability to multitask or focus on any task for a prolonged period is difficult in acute grief. Suggest that the family bring an additional family member or friend to important discussions to help them process and retain information and to serve as support.

*Last Modified April 2019*



- Be patient and understanding. Healing from emotional or psychological trauma takes time. Be patient with the pace of recovery and remember that there is no universal timeline for grieving. Don't judge one's reaction against your own response or anyone else's.
- Acknowledge the child's death by providing your sympathies to the family.
- Demonstrate that it is permissible to discuss the child who died and to express their emotions, including sadness, anger, and numbness. Remind them that it's ok to cry.
- Use the name of the child who died (do not refer to them as "the child who died"). Know their birth date and date of death and recognize the additional emotional impact they may have.
- Be honest and clear on what information is known and not known about the death of the child or risk to family members. Be clear about what is speculative and what is medical fact. Understand that the unexpected nature of the death may lead the family to develop their own theories and ideas. Listen to these respectfully and sensitively, while being clear about what is known and not known and providing directions for future investigation. Above all, remember that being an empathic listener and a calming presence is extremely helpful to these families.
- Assess their psychosocial support and coping skills and offer mental health support referrals as appropriate. Ask about sleep patterns, as grief is exhausting, and emotional resilience is compromised by poor sleep. Suggest that they discuss sleep problems with their primary care doctor. Educate families about the role of self-care (sleep, physical exercise, good nutrition, supportive friends/family) as important tools of self-efficacy and essential to their recovery. Be alert to signs and symptoms of:
  - Post-traumatic stress disorder (1 month or more): intrusive and distressing memories, avoidance that interferes with daily living, self-blame, feelings of detached detachment, hypervigilance
  - Complicated grief (6 months or more)-intense pining/longing for loved one, persistent rumination over the loss, problems accepting death, feeling that life no longer holds meaning or purpose
  - Depression- depressed mood, lack of interest, fatigue, suicidal thoughts

### 3. Meeting the Mental Health Needs of Family Members

Meeting and working with bereaved families can be difficult for clinicians. Many clinicians have not received formal training in providing any bereavement support, let alone the special care required after the sudden unexpected death of a child. An understanding of grief and trauma responses can be helpful in order to best meet the needs of families. Recognizing that families' needs may change over time and approaching all encounters with compassion and patience is of paramount importance to providing optimal care for families. It may be helpful for the clinician to seek consultation with colleagues around these challenging clinical situations.

When seeing a bereaved family member, it is important to remember that grief is normal, and that grief can manifest in a number of ways, including but not limited to regressive behavior, suicidal ideation, anxiety, depression, post-traumatic stress disorder (PTSD) and complicated grief (CG). Trauma can present with a wide range of physical, emotional and psychological symptoms.

*Last Modified April 2019*





Grief is a natural process of emotions that occurs when someone dies. Grief integrates into normal routines at variable time frames for each person. Complicated grief can occur when initial grief emotions continue and significantly impact functioning over 6 months or longer.

- a) Trauma can also be a component of grief when someone dies suddenly and unexpectedly, especially a child. This can leave survivors to feel helpless, vulnerable and often powerless. Trauma symptoms typically last from a few days to a few months, gradually fading as the emotional trauma is processed. Acceptance, processing the event and time are a part of the recovery process.

<b>Emotional and Psychological Symptoms of Trauma may include:</b> <ul style="list-style-type: none"> <li>• Shock, denial, or disbelief</li> <li>• Anger, irritability, mood swings</li> <li>• Guilt, shame, self-blame</li> <li>• Feeling sad or hopeless</li> <li>• Confusion, difficulty concentrating</li> <li>• Anxiety and fear</li> <li>• Withdrawing from others</li> <li>• Feeling disconnected or numb</li> <li>• Difficulty experiencing positive emotions</li> </ul>	<b>Physical Symptoms of Trauma may include:</b> <ul style="list-style-type: none"> <li>• Insomnia or nightmares</li> <li>• Flashbacks or other dissociative reactions</li> <li>• Being startled easily</li> <li>• Racing heartbeat</li> <li>• Aches and pains</li> <li>• Fatigue</li> <li>• Difficulty concentrating</li> <li>• Edginess and agitation</li> <li>• Muscle tension</li> </ul>
--	---

Gather information regarding patient's current coping mechanisms, support system, and mental health medical history. In office testing should be provided for at risk patients for possible referral to a psychiatrist or coordination with appropriate mental health services.

The following resources can be helpful in assessing a bereaved family member's grief response:

**American Psychological Association: Trauma**

<https://www.apa.org/topics/trauma/index.html>  
<https://www.apa.org/research/action/trauma>

**American Psychiatric Association: Post Traumatic Stress Disorder:**

<https://www.psychiatry.org/patients-families/ptsd/what-is-ptsd>

**Mayo Clinic: Complicated Grief:**

<http://www.mayoclinic.com/health/complicatedgrief/DS01023/DSECTION=symptoms>

**Complicated Grief Assessment**

[http://www.npcrc.org/files/news/complicated\\_grief\\_assessment.pdf](http://www.npcrc.org/files/news/complicated_grief_assessment.pdf)

**PHQ-9 to screen for depression**

<https://www.mdcalc.com/phq-9-patient-health-questionnaire-9>

Last Modified April 2019



## b) Therapeutic Interventions

Post-trauma reactions are normal and expected in the immediate aftermath of a sudden loss. It is important to normalize and not pathologize these reactions. Most individuals will experience natural recovery over the course of weeks or months, eventually regaining functioning in their lives. Resiliency is the norm.

However, some individuals will continue to struggle with persistent, post-trauma reactions (e.g., intrusive memories, avoidance of trauma reminders, intense negative emotions, hypervigilance) and may eventually develop posttraumatic stress disorder (PTSD). Individuals are at higher risk of developing PTSD if they lack positive social support. There are several effective, evidence-based psychotherapies for treating PTSD. The most commonly studied treatments are cognitive-behavioral therapies (CBTs) such as prolonged exposure (PE), and cognitive processing therapy (CPT). These CBTs focus on the interplay between thoughts, feelings, and behaviors in helping survivors to recover after traumatic events. Prolonged exposure (PE) specifically helps trauma survivors to gradually approach trauma-related memories, feelings, and situations they have been avoiding, allowing them to process the trauma and regain functioning in their lives. Cognitive processing therapy (CPT) specifically helps trauma survivors explore and change upsetting trauma-related thoughts and beliefs. CBTs are highly effective treatments for PTSD, associated with significant, lasting improvements in PTSD symptoms. Although working through trauma can feel scary at first, it is important to help trauma survivors understand that these treatments are not re-traumatizing; in fact, they can help survivors face their trauma, regain control over their lives and overcome debilitating PTSD symptoms.

EMDR (Eye Movement Desensitization and Reprocessing) is another psychotherapy for PTSD that incorporates elements of cognitive-behavioral therapy with eye movements or other forms of rhythmic, left-right stimulation. Medications, particularly antidepressants like SSRIs (selective serotonin reuptake inhibitors), are also effective in reducing PTSD symptoms.

### References:

- Asmundson, G. J., Thorisdottir, A. S., Roden-Foreman, J. W., Baird, S. O., Witcraft, S. M., Stein, A. T., ... & Powers, M. B. (2019). A meta-analytic review of cognitive processing therapy for adults with posttraumatic stress disorder. *Cognitive behaviour therapy*, 48(1), 1-14.
- Powers, M. B., Halpern, J. M., Ferenschak, M. P., Gillihan, S. J., & Foa, E. B. (2010). A meta-analytic review of prolonged exposure for posttraumatic stress disorder. *Clinical psychology review*, 30(6), 635-641.
- Watts, B. V., Schnurr, P. P., Mayo, L., Young-Xu, Y., Weeks, W. B., & Friedman, M. J. (2013). Meta-analysis of the efficacy of treatments for posttraumatic stress disorder. *J Clin Psychiatry*, 74(6), e541-e550.

Last Modified April 2019



**c) Recommendations on Supporting and Monitoring Siblings Experiencing Sudden Loss**

- Offer guidance to parents on age appropriate ways to talk to child about death
- Recommend age appropriate books (see Grief Resources link below)
- Make necessary mental health referrals: counselor, psychiatrist, support groups
- Consider making a flag or reminder in the child's chart
- Check in at regular intervals, particularly annual physicals, to see how child is coping, recognizing their needs may change over time
- Monitor for maladaptive responses that may necessitate more support or intervention: school difficulties, behavioral difficulties at home or school
- Recommend concrete way for them to remember their sibling: planting a tree, making a memory book, drawing pictures.
- Utilize the School Toolkit for siblings (see Grief Resources link below)
- Remember that siblings born subsequently may still experience grief and apply these strategies to them as well
- Remind families that familiar routines are very comforting to children and help them feel safe. For more specific information on supporting the needs of siblings, please visit the Grief Resources for Siblings page on the SUDC Foundation website.  
<https://sudc.org/education-literature/grief-resources/for-siblings>

**d) Professional support is recommended when the following are observed several months after the loss or trauma:**

- Trouble functioning at home or work
- Suffering from severe fear, anxiety, or depression
- Unable to form close, satisfying relationships
- Experiencing terrifying memories, nightmares, or flashbacks
- Avoiding things that are related to the trauma
- Feeling emotionally numb and disconnected from others
- Using alcohol or drugs to feel better

Educate the family on the various ways of coping and living after the death of a child. Acknowledge their grief and the fact that it is not linear and has no end point. Underscore how important it is for them to utilize resources to assist in finding productive ways to cope. There are countless ways for families to cope with their grief in a positive way, including but not limited to: participating in support groups, memorializing their child, fundraising for a cause to prevent future deaths, and pursuing legislative advocacy.

Making an early referral to the SUDC Foundation can help to reduce their isolation and connect them with other families who have experienced a similar loss. Referrals can be made to the SUDC Foundation before the autopsy is complete. If the death is sudden and

*Last Modified April 2019*





unexpected, the SUDC Foundation can provide resources, even if ultimately the death becomes explained.

e) Resources for meeting the emotional needs of the bereaved:

- National Center for Post-Traumatic Stress Disorder - <http://www.ptsd.va.gov>
- The Complicated Grief Program at Columbia University – <http://www.complicatedgrief.org/>
- American Psychiatric Association- [www.psychiatry.org](http://www.psychiatry.org)
- National Center for Post-Traumatic Stress Disorder - <http://www.ptsd.va.gov/>
- EMDR International Association (EMDRIA): Lists approved training programs, and a database of its certified members for client referrals.
- Scientific Working Group for Medicolegal Death Investigation's Principles for Communicating with Next of Kin during medicolegal Death Investigations. Published June 2012
- American Academy of Pediatrics: The Physician's Role When a Child Dies <https://www.aap.org/en-us/Documents/The%20Physician's%20Role%20When%20a%20Child%20Dies%20Discussion%20Guide.pdf>
- American Academy of Pediatrics Policy: Supporting the Family after the Death of a Child: <https://pediatrics.aappublications.org/content/130/6/1164>
- Center for the Study of Traumatic Stress: Helping People After a Loss: [https://www.cstsonline.org/assets/media/documents/CSTS\\_FS\\_Helping\\_People.pdf](https://www.cstsonline.org/assets/media/documents/CSTS_FS_Helping_People.pdf)

#### 4. Medical Care of Family Members after SUDC

Families who have experienced the sudden unexplained death of their baby or child often ask, "Should I have my other children tested?" This important question may be followed by the response of "Test them for what?" At this time, we do not know how to predict or prevent SUDC. However, we do know that some rare heritable disorders can cause sudden unexpected death. We also know that death investigations, as well as state mandated newborn screenings, vary across the U.S. Therefore, it is difficult to provide one specific set of tests for all families who have suffered an unexplained childhood death.

Research in the area of heritable disorders continues, and with the completion of the human genome project, new information is being attained. The ability to diagnose and treat individuals with these conditions continues to save lives and maximize the health of those affected. Without diagnosis and treatment, disorders such as Long QT Syndrome, various metabolic disorders, seizures and epilepsy disorders would continue to be the cause of sudden unexpected deaths in children and some adults.

Following optimal pediatric care guidelines for surviving and subsequent siblings is recommended. This includes attending well child visits, maintaining current vaccinations and following safe sleep guidelines in infancy (such as those of the AAP). The loss of a child affects the entire family dynamic. This loss needs to be taken into consideration as children's early environment and

Last Modified April 2019



experiences have crucial physical and mental effects on their lifelong health. Resources targeted toward health promotion and disease prevention should be made available.

In addition, the following information is to be considered with families who have suffered the sudden loss of a child where the cause of death is unknown, unclear or speculative. **The information is general and does NOT provide specific recommendations for any particular family** but provides a basis for discussions with families in order to determine appropriate screening tests, ensure accurate diagnoses and help provide some peace of mind for surviving family members in light of their specific needs, circumstance and medial information available. Specific evaluation by the family's physician will allow for the consideration of the below in addition to possibly other recommendations based on their review.

Collaboration between specially trained medical professionals is necessary to provide optimal care for the bereaved family of sudden child death. In order to develop an appropriate testing plan for current family members, and subsequent future pregnancies and children, it is important that **the primary care clinicians, the pathologist who performed the autopsy and any appropriate medical specialists discuss their specific recommendations with the affected family and each other.** These meetings may be conducted by the pathologist or the primary care physician, but it is crucial to have collaboration of the professionals involved to best meet the needs of the family.

**a) Recommended Follow Up from Primary Care Provider**

1. In the first 48 hours: Call the family upon receiving notification of death of child to express condolences and assess for necessary support
2. Send condolence card to family
3. Consider attending funeral services
4. Contact medical examiner to establish a timeline and determine how updates will be provided
5. Make referral to the SUDC Foundation for all sudden unexpected deaths in children. Even if a cause of death is determined, the SUDC Foundation will still provide services to families for any sudden child death.  
Referral Form: <https://sudc.org/direct-services/services-for-mdi-professionals>
6. 2 weeks after the child's death: call family and offer to have face to face meeting to discuss death of child/answer questions/meet with siblings/assess for need for referrals for support groups/mental health or medical referrals (cardiology/neurology/genetics).
7. Utilize the resources on the SUDC website to familiarize yourself with recommendations for screening for siblings and bereavement resources for families.
8. Connect the family with local bereavement support resources (if desired); for example, support groups through local churches and The Compassionate Friends and Bereaved Parents USA
9. Continue to contact family (if desired) at least monthly to assess for ongoing needs, recognizing that needs may change over time.
10. When the autopsy is complete, offer to read it with family. Offer to be present at a meeting with the medical examiner to explain the results.
11. Facilitate research opportunities for family, if desired, for more genetic testing/further evaluation.

*Last Modified April 2019*





12. Reach out to family on important dates for deceased child such as their birthday or anniversary of their death to express condolences and assess need for additional support.

#### **b) Considerations for Acute Care of SUDC Siblings**

When an otherwise healthy child dies, and the medical community can offer no answers, parents will understandably be more anxious regarding their surviving and subsequent children. This may prompt more visits to their pediatricians or emergency rooms for what may otherwise appear to be a mild illness. It is important to find a balance between providing sensitive care to families in these interactions, while also not over medicalizing the sibling.

#### **References:**

Julie P. Katkin, Susan J. Kressly, Anne R. Edwards, James M. Perrin, Colleen A. Kraft, Julia E. Richerson, Joel S. Tieder, Liz Wall, TASK FORCE ON PEDIATRIC PRACTICE CHANGE; American Academy of Pediatrics Policy Statement Guiding Principles for Team-Based Pediatric Care Pediatrics August 2017, VOLUME 140 /ISSUE 2

Rogers CH, Floyd FJ, Seltzer MM, Greenberg J, Hong J. Long-term effects of the death of a child on parents' adjustment in midlife. *J Fam Psychol.* 2008;22(2):203–211.  
doi:10.1037/0893-3200.22.2.203

Crandall L, Devinsky O. Sudden Unexplained Death In Children. *The Lancet Child and Adolescent Health*, Vol 1, September 2017

#### **c) DNA Banking**

Currently, genetic analysis of the SUDC child is not routine across the U.S. during a medicolegal death investigation, even though recent research has shown potentially causative variants in up to 20% of the SIDS cases. Genetic research into SUDC is in the early stages but have identified both cardiac and neurological pathogenic variants in victims.

The SUDC Foundation does provide assistance in DNA banking as a precursor to genetic testing referrals for families. DNA banking early on in the investigation, with specimens retained at the autopsy, allows for the preservation of DNA from the child to ensure opportunities in the future when the family may be interested in research and/or clinical opportunities to attain genetic analysis of their child.

DNA banking early on is also important as storage times for specimens at medical examiner and coroner offices vary widely. For more information on **DNA banking services at the SUDC Foundation**, go to: [sudc.org](http://sudc.org) > **medical information** > **DNA banking**

#### **References:**

Halvorsen M, Petrovski S, Shellhaas R, Tang Y, Crandall L, Goldstein D, Devinsky O.

*Last Modified April 2019*



Mosaic mutations in early-onset genetic diseases Genet Med. 2016 Jul;18(7):746-9. doi: 10.1038/gim.2015.155. Epub 2015 Dec 30. PMID: 26716362 Free PMC Article available

Neubauer J1, Lecca MR2, Russo G2, Bartsch C3, Medeiros-Domingo A4, Berger W5,6,7, Haas C1. Post-mortem whole-exome analysis in a large sudden infant death syndrome cohort with a focus on cardiovascular and metabolic genetic diseases. Eur J Hum Genet. 2017 Apr;25(4):404-409. doi: 10.1038/ejhg.2016.199. Epub 2017 Jan 11.

#### d) Evaluating for Inborn Errors of Metabolism

Some questions to consider:

- What was the extent and result of the state mandated newborn screening?
- Is the newborn card available now for retrieval?
- Did the child who died have any symptoms that could be suggestive of metabolic disease, such as vomiting, decreased appetite, lethargy, febrile illness, etc. in the days preceding their death?
- Did the pathologist discover autopsy findings, medical history findings or terminal history findings that raise the concern for metabolic disease?
- What was the extent and result of post mortem metabolic studies performed on the child who died? (Resource: [www.savebabies.org](http://www.savebabies.org))

#### Evaluating Siblings:

##### During the PRENATAL period:

- Patient may want to consider genetic counseling or preconception genetic screening.
- Encourage patient to get regular and early prenatal care.
- Special consideration should be given to the possible occurrence of maternal complications: Acute Fatty Liver of Pregnancy (AFLP), Pre-Eclampsia, either mild, which is more common, or severe manifestation as HELLP (hemolysis, elevated liver enzymes, low platelets)

##### For the Asymptomatic SUDC/SIDS/SUID Sibling NEWBORN:

- Blood: Expanded newborn screening by MS/MS (blood spots)—If this is not mandated in your state, bloodspot cards can be requested ahead of time from private labs. (Information on some available labs can be obtained from the SUDC Foundation)
- Urine: Organic Acids
- Consider repeating above at time of first illness

##### For the Symptomatic SUDC/SIDS/SUID Sibling who is a NEWBORN:

- If the newborn is not well—Seek emergency medical attention. Consult with specialists in metabolic disease.
- Start evaluation with investigations that include:
- Blood Gases, Glucose, Electrolytes, Lactate, Pyruvate, Ammonia, Urine Organic Acids

Last Modified April 2019



For Symptomatic SUDC/SIDS/SUID Older Siblings:

- PLASMA Acylcarnitines and Urine Organic Acids
- If expanded newborn screening, plasma acylcarnitines and organic acids are all negative, the pursuit of additional targeted tests should be considered on a case-by-case basis, also determined by the outcome of the postmortem investigation in the child who died suddenly.
- Consider repeating above during times of illness.

*References:*

Yamamoto T1 et al. Metabolic autopsy with next generation sequencing in sudden unexpected death in infancy: Postmortem diagnosis of fatty acid oxidation disorders. *Mol Genet Metab Rep*. 2015 Oct 2; 5:26-32. doi: 10.1016/j.ymgmr.2015.09.005. eCollection 2015 Dec.

Rinaldo P, Matern D, Bennett MJ. Fatty acid oxidation disorders. *Ann Rev Physiol* 64:16.1-26, 2002.

Bennett MJ, Rinaldo P. The Metabolic Autopsy comes of Age. *Clinical Chemistry* 47; 1145-6, 2001

Tortorelli S, Hahn SH, Cowan TM, Brewster TG, Rinaldo P, Matern D. The urinary excretion of glutarylcarnitine is an informative tool in biochemical diagnosis of glutaric acidemia type 1. *Molecular Genetics and Metabolism* 84:137-143, 2005  
Mayo Medical Laboratories Communiqué on postmortem testing

**e) Evaluating for Cardiac Channelopathies**

In addition to discussion of the past medical history of the child who died, their post mortem findings and the family's medical history, consider these specific questions:

- Is there a family history of Sudden Death in those under 50 years of age?
- Is there a family history of Unexplained Syncope, Sleep disorders, ALTE or Seizures?
- Is there a history of Syncope, Sleep disorders, ALTE, Seizures in the child who died?

Although heart muscle and congenital abnormalities, tumors and infection can be evaluated during the standard autopsy procedures, some cardiac abnormalities require a molecular autopsy and/or careful examination of surviving family members for full evaluation.

1. Therefore, in 2011, the HRS/EHRA Expert Consensus Statement on the State of Genetic Testing for Channelopathies and Cardiomyopathies published in *Heart Rhythm*, Vol 8, No 8, August 2011 provided recommendations on the "STATE OF POST-MORTEM GENETIC TESTING IN SUDDEN UNEXPECTED DEATH CASES (SUD/SIDS)" The Expert Consensus Recommendations concluded: For all SUDS and SIDS cases, collection of a tissue sample is recommended (5–10 mL whole blood in EDTA tube, blood spot card, or a frozen sample of heart, liver, or spleen) for subsequent DNA analysis/genetic testing. In the setting of autopsy-negative SUDS, comprehensive or targeted ion channel genetic testing may be

*Last Modified April 2019*





considered in an attempt to establish probable cause and manner of death and to facilitate the identification of potentially at-risk relatives and is recommended if circumstantial evidence points toward a clinical diagnosis of Long QT Syndrome or Catecholaminergic Polymorphic Ventricular Tachycardia (CPVT) specifically (such as emotional stress, acoustic trigger, drowning as the trigger of death).

2. Mutation-specific genetic testing is recommended for family members and other appropriate relatives following the identification of a SUDS-causative mutation in the decedent. Discussions regarding genetic testing of Cardiac Channelopathies among families and their physicians need to consider what genetic samples are available for testing from the child who died.

#### **Channelopathy Screening of the Child who Died**

- Options through research and commercial testing- see [sudc.org](http://sudc.org)
- Genetic testing via research (no cost to families for genetic testing of child and both parents):



Review and refer families at: [sudc.org](http://sudc.org) > research and medical information > SUDCRRRC

Any positive genetic finding requires detailed analysis to determine if the identified mutation is truly disease-causing or a benign variant. This can be supported by clinical and genetic evaluation of surviving family members.

#### **Channelopathy Screening of Family Members**

Since channelopathies can be inherited, other biological relatives may be at risk. Therefore, evaluation of family members for long QT Syndrome and other inherited arrhythmia conditions is appropriate to both confirm a family diagnosis and assess the risk in the living family members following an unexplained death or a sudden cardiac death. Clinical evaluation of family members following a sudden death may identify other affected members and hence provide a diagnosis in up to 40% of families (Ref Tan) and may be supported by any genetic findings.

First degree relatives (parents and siblings of the person who has died) should speak to their primary care physician to obtain a referral to a cardiologist (with expertise in electrophysiology and special training to evaluate genetic cardiac disorders) to perform:

1. A VERY careful and detailed medical history of the decedent with review of the autopsy report and sometimes the autopsy tissue itself by a cardiac pathologist.
2. A VERY careful and thorough review of the family history with pedigree analysis on both sides searching for relevant history or events on either side of the family. Current research has found a greater incidence of channelopathies diagnosed postmortem with the greater age of the child. Therefore, in older children where the autopsy fails to demonstrate any structural abnormalities, a clinical screen should be

*Last Modified April 2019*



performed in surviving family members to assess for the possibility of inherited heart rhythm syndromes.

- Clinical screening tests would include: a. ECG/EKG, b. Holter, and c. Exercise Stress Test
- (Exercise stress test recommendation is based on the findings and recommendations of the 3rd reference below, due to CPVT escaping detection by a resting electrocardiogram).
- Any additional provocative tests (i.e. epinephrine QT stress test, procainamide stress test, or isoproterenol stress test, or diagnostic EP study) would be based upon the clinical impression, family history, etc.
- If directed by results of such investigations, molecular genetic testing for a "cardiac channelopathy" like long QT syndrome should be pursued.

#### Locating a Specialist for Cardiogenetic Screenings:

Online resources to find a specialist in your area include:

- The Pediatric and Congenital Electrophysiology Society  
<http://pediatricpsociety.org/>
- The SADS Foundation (Sudden Arrhythmia Death Syndromes)  
<https://www.sads.org/>
- Some cardiogenetic clinics are also listed on the SUDC Foundation website at:  
[sudc.org](http://sudc.org) > medical information > medical care after SUDC

If you are having trouble locating a specialist, please contact the SUDC Foundation (toll free in the U.S) at 800-620-SUDC (7832) or direct dial at 973-795-1257.

#### References:

Ackerman et al. HRS/EHRA Expert Consensus Statement on the State of Genetic Testing for Channelopathies and Cardiomyopathies. Heart Rhythm, Vol 8, No 8, August 2011

Bagnall RD et al , Weintraub RG, Ingles J, Duflo J, Yeates L, Lam L, Davis AM, Thompson T, Connell V, Wallace J, Naylor C, Crawford J, Love DR, Hallam L, White J, Lawrence C, Lynch M, Morgan N, James P, du Sart D, Puranik R, Langlois N, Vohra J, Winship I, Atherton J, McGaughan J, Skinner JR, Semsarian C. A Prospective Study of Sudden Cardiac Death Among Children and Young Adults. N Engl J Med. 2016 Jun 23;374(25):2441-52. doi: 10.1056/NEJMoa1510687. PMID: 27332903

Ackerman MJ, Tester DJ, Driscoll, DJ. Molecular autopsy of sudden unexplained death in the young. The American Journal of Forensic Medicine and Pathology. 2001; 22(2): 105-111.

Ackerman MJ, Khosiseh A, Tester DJ, Hejlik J, Shen WK, Porter CJ. Epinephrine-induced QT interval prolongation: a gene-specific paradoxical response in congenital long QT syndrome. Mayo Clinic Proceedings. 2002;77:413-421.

Tester DJ, Spoon DS, Valdivia HH, Makielski JC, Ackerman MJ. Targeted Mutational Analysis of RyR2-Encoded Cardiac Ryanodine Receptor in Sudden Unexplained Death: A

*Last Modified April 2019*

The SUDC Foundation ★ 101 Eisenhower Pkwy, Ste 300 ★ Roseland, NJ 07068



Molecular Autopsy of 49 Medical Examiner/Coroner Cases. Mayo Clinical Proceedings, 2004, 79 (11): 1380-1384 [www.mayo.edu/proceeding](http://www.mayo.edu/proceeding)

Tan H, Hofman N, Van Langen IM, Van Der Wall AC, Wilde AAM. Sudden unexplained death: heritability and diagnostic yield of cardiological and genetic examination in surviving relatives. Circulation 2005;112: 207-213.

#### **f) Evaluating Seizures in Siblings of SUDC**

Families registered with the SUDC Foundation are reporting a higher than expected incidence of seizures, especially febrile seizures, in both the child who died and sometimes in other family members as well. Medical literature over the last several decades reports on the benign nature of simple febrile seizures. At this time, we recommend a consultation with a pediatric neurologist for all SUDC siblings who have a history of seizures (febrile or without fever) or any other neurological concern (e.g., developmental delay).

The articles below may provide some information and guidance to families and physicians. At this time, since the association between SUDC and febrile seizures is still being investigated, we cannot make any general recommendations about the specific evaluation and treatment of febrile seizures in siblings of children who have died of SUDC.

If, at any time, contact with The SUDC Foundation for more information and/or direct contact with our researchers or physicians would be helpful in evaluating these children, please call Laura Crandall toll free in the U.S. at 800-620-SUDC (7832) or direct dial 973-795-1257.

#### **References:**

Crandall L, Devinsky O. Sudden Unexplained Death In Children. The Lancet Child and Adolescent Health, Vol 1, September 2017

Hesdorffer DC, Crandall LA, Friedman D, Devinsky O; Sudden unexplained death in childhood: A comparison of cases with and without a febrile seizure history. Epilepsia. 2015 Aug;56(8):1294-300. doi: 10.1111/epi.13066. Epub 2015 Jun 29

Hefti MM, Kinney HC, Cryan JB, Haas EA, Chadwick AE, Crandall LA, Trachtenberg FL, Armstrong DD, Grafe M, Krous HF. Sudden unexpected death in early childhood: general observations in a series of 151 cases: Part 1 of the investigations of the San Diego SUDC Research Project Forensic Sci Med Pathol. 2016 Mar;12(1):4-13. doi: 10.1007/s12024-015-9724-2. Epub 2016 Jan 19. PMID: 26782961 Free PMC Article

Ingrid A. Holm MD, MPH, Annapurna Poduri MD, MPH, Laura Crandall PT, MA, Elisabeth Haas MPH, Marjorie R. Grafe MD, PhD, Hannah C. Kinney MD, Henry F. Krous MD, Inheritance of Febrile Seizures in Sudden Unexplained Death in Toddlers. Pediatric Neurology 46 (2012) 235e239

Mazumdar, M Febrile Seizures and Risk of Death (editorial) Lancet. 2008; Vol 372:429-430. Krous HF, Chadwick AE, Crandall L, Nadeau-Manning. Sudden Unexpected Death In Childhood: A Report of the 50 Cases. Pediatric Dev Pathol. 2005 July 14

*Last Modified April 2019*

[www.sudc.org](http://www.sudc.org) ★ 800.620.SUDC(7832) ★ Direct 973.795.1257



The SUDC Foundation ★ 101 Eisenhower Pkwy, Ste 300 ★ Roseland, NJ 07068



5. \*\*\*Note to Parents affected by SUDC\*\*\*

With continued research, we hope to further refine this information to provide the best care and hope for the future of all families. The SUDC Foundation offers the above information as a starting point for discussions with your personal doctor, who knows your family best.

Discuss the above with your doctor. Ask him/her to evaluate this information in the context of the specific outcome of the postmortem investigation of the child who died suddenly and any pertinent family history. If additional clarification or assistance is needed for understanding this document, please contact The SUDC Foundation for the direct contact information of the contributing professionals.

**Family services are available for you at no cost through the SUDC Foundation. Contact us at [info@sudc.org](mailto:info@sudc.org), call 800-620-SUDC or register yourself at [www.sudc.org](http://www.sudc.org).**

**For More information on SUDC: [www.sudc.org](http://www.sudc.org)**

*The SUDC Foundation is grateful to our Scientific Advisory Board and Medical Education Committee for their assistance in the creation of this resource.*

*Please refer to [sudc.org](http://sudc.org) to ensure most current version of this document.*

*Last Modified April 2019*

[www.sudc.org](http://www.sudc.org) ★ 800.620.SUDC(7832) ★ Direct 973.795.1257