

University of Warwick institutional repository: <http://go.warwick.ac.uk/wrap>

This paper is made available online in accordance with publisher policies. Please scroll down to view the document itself. Please refer to the repository record for this item and our policy information available from the repository home page for further information.

To see the final version of this paper please visit the publisher's website. access to the published version may require a subscription.

Author(s): Peter Sidebotham

Article Title: Unexpected but not unexplained: investigating a case of sudden unexpected death in infancy

Year of publication: 2005

Link to published version: <http://dx.doi.org/10.1136/adc.2004.057430>

Publisher statement: None



Unexpected but not unexplained: investigating a case of sudden unexpected death in infancy

Peter Sidebotham

Arch. Dis. Child. Ed. Pract. 2005;90;ep40-ep45
doi:10.1136/adc.2004.057430

Updated information and services can be found at:
<http://ep.bmjournals.com/cgi/content/full/90/2/ep40>

These include:

References

This article cites 9 articles, 4 of which can be accessed free at:
<http://ep.bmjournals.com/cgi/content/full/90/2/ep40#BIBL>

Rapid responses

One rapid response has been posted to this article, which you can access for free at:

<http://ep.bmjournals.com/cgi/content/full/90/2/ep40#responses>

You can respond to this article at:

<http://ep.bmjournals.com/cgi/eletter-submit/90/2/ep40>

Email alerting service

Receive free email alerts when new articles cite this article - sign up in the box at the top right corner of the article

Notes

To order reprints of this article go to:

<http://journals.bmj.com/cgi/reprintform>

To subscribe to *Archives of Disease in Childhood - Education and Practice* go to:

<http://journals.bmj.com/subscriptions/>

UNEXPECTED BUT NOT UNEXPLAINED: INVESTIGATING A CASE OF SUDDEN UNEXPECTED DEATH IN INFANCY

ep40

Peter Sidebotham

Arch Dis Child Educ Pract Ed 2005;90:ep40–ep45. doi: 10.1136/adc.2004.057430

A 10 day old boy called Jordan was brought into the accident and emergency department by ambulance at 4.30 am one Sunday morning in October in a collapsed state. His mother had called for an ambulance 20 minutes earlier. When the paramedic crew arrived at the house they found a pale, lifeless infant with no pulse and no respiratory effort. They had instituted cardiopulmonary resuscitation using a bag valve and mask and continued this during the transfer to hospital. His mother accompanied him in the ambulance.

The arrest team had been alerted before arrival and was on hand in the department. The paediatric registrar made some initial preparations and requested that the on-call consultant be notified. Immediate evaluation confirmed the absence of a pulse and respiratory effort. Full cardiopulmonary resuscitation was instituted, including tracheal intubation and ventilation, and insertion of an intraosseous line with administration of adrenaline and a fluid bolus. On the cardiac monitor the ECG showed a flat trace. The rectal temperature was 35.1°C. A BM stick taken at insertion of an intravenous line showed a blood sugar concentration of 2.0 mmol/l. There was insufficient blood for blood gas analysis or other investigations, but the syringe was flushed with medium from a blood culture bottle. The mother, and shortly after the maternal grandmother, were present in the resuscitation room with the staff nurse assigned to support them.

Since the team had received a brief warning of the emergency, they had been able to make some preparations with the allocation of roles and the preparation of appropriate drugs and equipment. All the team members had attended paediatric life support training and this facilitated a good team approach and a prompt, appropriate, and un-panicked resuscitation. This included the designation of a nurse to support the mother and grandmother. This nurse also gave the mother the option of being present during the resuscitation.

The priority was initially to attempt resuscitation. At the same time the paediatric registrar was quickly considering the differential diagnosis. Highest on his list were infection and congenital heart disease, and in addition an inborn error in metabolism and non-accidental injury crossed his mind. Since there were no signs of life he also considered sudden infant death syndrome. In view of the difficulty in obtaining intravascular access and blood samples he was forced to prioritise the investigations, guided by an appreciation of the most likely and most treatable conditions.

COMMENT

- ▶ Most cases of sudden collapse of an infant will present at hospital following an emergency telephone call. Unless the infant has clearly been dead for some time (with signs of rigor mortis), resuscitation should be attempted until arrival in hospital. It should then be continued according to the Advanced Paediatric Life Support Protocol¹ until there has been a thorough appraisal of the situation, taking account of the clinical features, the response to appropriate resuscitation, and any immediately available results of investigations.
- ▶ Even where the infant has clearly been dead for some time, it is helpful for all cases of sudden unexpected death in infancy (SUDI) to be brought to hospital. This serves a number of important purposes. Firstly, a fully equipped team will be on hand to thoroughly evaluate the situation; secondly, the team will be able to support the parent(s) and other family members; thirdly, it helps to secure the scene of death which can then be thoroughly evaluated later.
- ▶ During any resuscitation, blood should be taken promptly for glucose and electrolyte assay and for bacteriological culture. However, after death has been pronounced no samples may be taken from the baby without the consent of the coroner. Providing a necropsy is performed within 24 hours of death, most investigations can be done at that time; however, bacteriological samples may be affected by any delay and should be taken at the time with the agreement of the coroner. The coroner is in charge of the situation, but most will agree to a standard approach to investigation thereby avoiding the need for specific consent in every case. Any samples should be carefully labelled and stored in case further analysis is required. If there is a history suggestive of possible metabolic disorder, an early frozen section of liver can be

Correspondence to:
Dr Peter Sidebotham,
Division of Health in the
Community, University of
Warwick, Coventry CV4 7AL,
UK; p.sidebotham@warwick.ac.uk

invaluable. A protocol for the immediate investigation of SUDI is provided in the Kennedy report (table 1).²

- ▶ A low blood glucose is a common and non-specific finding in paediatric arrest and in any infant who is acutely unwell, stressed, or hypoxic. Hypoglycaemia may arise as a result of sepsis, poisoning, hypothermia, metabolic disorders, and asphyxia. Steps should be taken during resuscitation to correct any underlying hypoglycaemia.¹
- ▶ The rectal temperature of 35.1°C is in keeping with the death having occurred within a few hours of presentation. There have been numerous attempts to describe methods of estimating the time of death from the deep body temperature, mostly based on adult studies.³ Postmortem body cooling will be influenced by a large number of factors including body weight, ambient temperature, body temperature at the time of death, and clothing. The rectal temperature cannot be used as a pointer to any specific cause of death.

Resuscitation in hospital was continued for 20 minutes but since there was still no response it was decided to stop. The registrar carefully explained this to Jordan's mother and grandmother. The intraosseous line was removed, and after directly visualising the position of the tracheal tube, this was also removed. Jordan was then wrapped in a baby blanket and given to his mother to hold. Jordan's mother and grandmother were given time on their own with Jordan in a small private room, supported by the staff nurse.

Meanwhile, the paediatric consultant arrived and was briefed by the registrar. Uniformed police officers (alerted by ambulance control) also arrived in the department, and following discussion with the consultant they agreed to contact the police child protection team. The consultant then introduced himself to the mother and obtained a basic history and briefly reviewed what had happened. He then explained the process of investigation. Two members of the police child protection team joined him during this history

taking and explained their role in the overall management. The consultant explained the scope of the investigation of Jordan's death including the involvement of the coroner, the need for a postmortem examination, and arranged a home visit for later that day. He took Jordan back into the resuscitation room for a full examination and for mementos to be taken, including hand and foot prints, a photograph, and a lock of hair. The staff nurse provided Jordan's mother with an information pack produced by the Foundation for the Study of Infant Deaths (FSID) (www.sids.org.uk) and the contact numbers of the clinical coordinator and the FSID parents' helpline.

A telephone discussion was then held between the consultant paediatrician, the police officers, and the emergency duty social worker to confirm basic details, including details of all family and household members, and to plan the investigation.

Having decided to stop resuscitation, the team was now dealing with a case of SUDI. At this stage there were no clear indicators of why Jordan had died. Nevertheless, the fact that Jordan was only 10 days old was not typical of sudden infant death syndrome (SIDS). The consultant kept this in mind in his discussions with the other agencies and in his approach to the further investigation, the first part of which was to obtain a full history (table 2). Jordan's age made it much more likely that there was an underlying medical or forensic reason for the death.

COMMENTS

- ▶ As Baroness Kennedy pointed out in her recent report on SUDI, it is every family's right to have their baby's death properly investigated.² This can only fully be achieved through a clear multi-agency approach to investigation and family support.²⁻⁴ The first stage of such an approach is a strategic discussion involving the paediatrician, police, and social services. This enables sharing of information, identification of any initial concerns, a clear division of

Table 1 Initial samples to be taken immediately after sudden unexpected death in infancy (SUDI)²

Sample	Send to:	Handling	Test
Blood (serum) 0.5 ml	Clinical chemistry	Normal	Urea and electrolytes
Blood (serum) 1 ml	Clinical chemistry	Spin, store serum at -20°C	Toxicology
Blood (lithium heparin) 1 ml	Clinical chemistry	Spin, store plasma at -20°C	Inherited metabolic diseases
Blood (fluoride) 1 ml	Clinical chemistry	Spin, store plasma at -20°C	3-OH butyrate, sugar, FFA, lactate
Blood EDTA 0.5 ml	Haematology	Normal	FBC
Blood cultures— aerobic and anaerobic 1 ml	Microbiology	If insufficient blood, aerobic only	Culture and sensitivity
Blood from syringe onto Guthrie card	Clinical chemistry	Normal (fill in card—do not put into plastic bag)	Inherited metabolic diseases
Blood (lithium heparin) 5 ml	Cytogenetics	Normal—keep unseparated	Chromosomes (if dysmorphic)
CSF (a few drops)	Microbiology	Normal	Microscopy, culture and sensitivity
CSF 0.5 ml	Clinical chemistry	Store at -20°C	Inherited metabolic diseases
Swabs from any identifiable lesions	Microbiology	Normal	Culture and sensitivity
1. Urine (if available)	Clinical chemistry	Spin, store supernatant -20°C	Toxicology, inherited metabolic diseases
2. Urine (few drops, if above sample taken)	Microbiology	Normal	Microscopy, culture and sensitivity

Any postmortem investigations need to be agreed with the coroner. Often this can be agreed in advance as a standard protocol. Most investigations can safely be done at necropsy. CSF, cerebrospinal fluid; EDTA, ethylene diamine tetraacetic acid; FBC, full blood count; FFA, free fatty acid; OH, hydroxy.

Table 2 A structured clinical history in SUDI

- ▶ Basic details of baby, the parents, and other family members
- ▶ A narrative account of the 24 hours leading up to the baby's death, including a full description of when and how the baby slept and fed, any activity, who was with the baby at different times, the baby's health and activity levels, and any changes to routine
- ▶ The final sleep
- ▶ Where and how the baby was put down, clothing, bed coverings, position; any changes in that during the course of the night; if bed sharing, who else was in the bed and their positions relative to the baby; when and by whom the baby was checked during the sleep; description of the last feed and any night time feeds; heating and ventilation
- ▶ Where and how the baby was found, position, coverings, appearance and any unusual features; any action taken after the baby was found
- ▶ Baby's past medical history, including pregnancy and delivery, growth and development, feeding, any illnesses, immunisations and routine surveillance; also details of normal routine for the baby, including feeding sleeping patterns and practices
- ▶ Family medical history, including any medical or psychiatric history of the parents and other immediate family members; infectious contacts; any history of respiratory, cardiac, or neurological disorders in the family and any previous infant or other sudden deaths in the family
- ▶ Social history
- ▶ Family structure and dynamics, housing, use of alcohol, recreational drugs, and tobacco; parents' occupations; any social services involvement in the past, including any child protection concerns

responsibilities, and a plan for investigating the case and for giving information to and supporting the family. At this stage, or at any stage if distinct child protection concerns are identified, a formal strategy meeting should be convened by social services under section 47 of the Children Act.

- ▶ A consultant paediatrician should take the lead in coordinating the investigation and family support following a sudden infant death. This may be the consultant who initially sees the family in the emergency department, or may be passed on to a community paediatrician or a designated SUDI paediatrician. If the case is transferred from one consultant to another there should be a clear handover of responsibilities. This paediatrician will have to work very closely with the police child protection team. As far as possible, repeated questioning of the parents should be avoided, and this can often be achieved by joint interviews involving both the police and the paediatrician, either at home or in the emergency department. This, however, will not always be practical and there may well be situations where the police will nevertheless need to interview the parents separately.
- ▶ It is important to use the appropriate terminology. At these early stages, any unexpected death should be referred to as a sudden unexpected death in infancy. This is a descriptive term, and does not imply causation. Many such deaths will turn out to have an identified cause, either medical or forensic, but that can only be determined after a thorough investigation including a full, structured history, an examination of the scene, and a comprehensive postmortem examination. The term "sudden infant death syndrome" should be reserved for the sudden death of an infant under 1 year of age that remains unexplained after a meticulous investigation, including a thorough necropsy, examination of the death scene, and review of the clinical history.⁵ At least 20% of SUDI will have an identifiable medical cause, such as infection, metabolic disorder, or previously unrecognised congenital malformations; up to 10% may be a result of homicide; and the remainder will be attributed to SIDS.⁶

- ▶ Since the campaigns to reduce the risk of SIDS in the early 1990s, the overall incidence of SUDI has dropped from around 2 per 1000 livebirths in the 1980s to 0.48 per 1000 in 2002.⁷ Nevertheless, SIDS remains the largest single cause of death in the post-neonatal period. A number of distinct epidemiological characteristics have been identified.⁵ Most deaths occur between 6 weeks and 6 months of age and there is an increased incidence after preterm birth, in low birthweight infants, and in males. Young, single mothers, and those from more deprived backgrounds are more at risk. There is a clear association with parental smoking and with placing babies to sleep in the prone position. There have been a lot of theories on the pathogenesis of SIDS, but overall it remains unexplained. The most widely accepted hypothesis posits that "the fatal event involves a neurally-compromised infant, circumstances that challenge vital physiology, most likely during sleep, at a particular developmental period".⁸
- ▶ One of the most interesting findings to come out of epidemiological research is that many of the risk factors are shared between SIDS, explained SUDI, and deaths caused by maltreatment. These risk factors, including indicators of social deprivation, may be markers of genetic influences transmitted from one generation to another, or may act through effects on parenting, nutrition, or environmental stress.
- ▶ One of the most important aspects of any SUDI investigation is to treat the baby and the family with sensitivity and respect. The removal of cannulae, catheters, and tracheal tubes is appropriate unless they may have contributed to the death. Before removing a tracheal tube, its position should be confirmed by direct visualisation, ideally by someone other than the doctor who carried out the intubation. A complete record of all cannulation attempts, etc, monitors and tubes should be made before their removal. Always refer to the baby by name. Provide ample time for the parents to be with and hold their baby (this will not compromise any investigation and should be discussed with the police team). Take mementos that can be given to the parents if they so wish.

The consultant took an initial history in the department and found out that Jordan was the third child of parents who were now separated. The two older children were living with the paternal grandparents following a court order made because of neglect. Jordan's mother, Emma, had a longstanding history of substance abuse. She had been on a methadone programme (and off other drugs) for most of the pregnancy and since, supervised by the local drug rehabilitation team. Jordan had been born at 36 weeks gestation with a birthweight of 2620 g. He had shown some signs of drug withdrawal, but had not required any treatment. He was initially tube fed, but normal feeding was quickly established and he was discharged along with his mother to the maternal grandmother's home at 8 days of age. Emma admitted to smoking 10–20 cigarettes per day. She had taken her methadone as usual the previous day and had consumed at least three units of alcohol that evening.

Jordan had remained well following discharge from hospital. On the day before his death he had fed normally. Emma had given him a bath and put him to bed at about 7 pm. She had herself gone to bed at around 11 pm and had not noticed anything worrying about Jordan. She then went to sleep with Jordan beside her on the bed. She woke up in the early hours

of the morning feeling something was wrong. She immediately noticed that Jordan was not making any sound, and that the blanket was partly covering his face. When she picked him up he was floppy and lifeless. Emma had immediately gone in to her mother's room and they telephoned for an ambulance.

At this stage major concerns were beginning to emerge about Emma's parenting ability, with the history of substance abuse, previous neglect of her children, and parental separation. It was recognised that these aspects, together with the slight prematurity, low birthweight, and neonatal abstinence syndrome, had made Jordan very vulnerable. At 10 days of age, he was outside the typical age profile for SIDS. The consultant was also concerned about the co-sleeping and whether that had contributed to death. It was agreed that a more formal strategy meeting with professionals who knew the family was needed to review the details of these concerns and plan further investigations. This meeting would also consider the support Emma might need, recognising that she was in a particularly vulnerable situation.

COMMENTS

► As in most aspects of medical practice, the taking of a thorough systematic history is of crucial importance in the evaluation of cases of SUDI. As well as giving pointers to possible underlying medical disorders—for example, through factors in the previous medical or family history—a thorough history may help to elucidate factors in the parents' knowledge, parenting practices (for example, smoking or sleeping position), or social situation which, while not causal, may have contributed in part to the death, and may be amenable to change in future pregnancies; or may identify discrepancies, or concerning information that warrants further detailed investigation.

The consultant convened a strategy meeting which was held at the general practitioner's (GP's) surgery later that morning and involved the police officers, social worker, GP, and midwife. This was followed by a joint home visit by the consultant, a police officer, and the GP. During this visit, a more detailed history was obtained, including a review of Emma's history, her pregnancy, the wider family history, and a step by step account of the 24 hours leading up to Jordan's death. The police officer and paediatrician then examined the room, looking at the bedding and sleeping arrangements.

Emma had been sleeping with Jordan in a single bed in a small bedroom. She had put Jordan down on his back on her bed as normal. He had been wearing a vest and a baby gro, but no hat or gloves. She had wrapped him in a cot blanket and covered him with another small cot blanket. It had not been a particularly cold night, but the door and windows had been closed. There were no unusual findings on the scene examination.

It transpired that the previous children had been removed because of concerns about neglect relating to both parents' drug habits and their unstable relationship. A case conference had been held before Jordan's birth, and it had been agreed to allow Jordan to stay with his mother, given that she was cooperating with

the methadone programme, had separated from her partner, and was living with her mother who was able to provide support and extra care. The midwife felt that Emma might have been showing some early signs of postnatal depression and she had been intending to discuss this with the GP.

The social situation seemed more stable than had been originally thought and there was nothing specific in the history or scene examination, yet Jordan's death was still unexplained and the possibility of homicide remained. Emma was kept informed that as in all cases of SUDI, a full investigation including the postmortem examination is needed before any indication of the cause of death can be given. Because the paediatrician had taken a thorough history in the presence of the police officers, there was no need at this stage for them to take a separate statement from Emma. She was told that the team was liaising with her social worker and midwife to both identify any factors that may have contributed to Jordan's death and to ensure their continued support of her. The basic details regarding what a postmortem examination involves were explained.

COMMENT

► Although the vast majority of SUDI cases arise from natural (though not necessarily well understood) causes, a significant proportion occur as a direct or indirect result of child maltreatment. Estimates of the proportion of SUDI caused by homicide vary considerably⁹ although many authors suggest up to 5–10% may be frank homicide, with maltreatment (abuse or neglect) being a contributory (though not necessarily causal) factor in a similar proportion. Maltreatment associated deaths may result from severe non-accidental injury including cases of shaking or shaking-impact injuries; from extreme neglect, or from induced illness as a result of, for example, poisoning or asphyxiation. Deaths from suffocation may be particularly difficult to detect, as there may be no specific features in the history, scene examination, or post-mortem examination. Certain factors may increase suspicions regarding maltreatment¹⁰ (table 3), although it is important to recognise that none of these findings are diagnostic and in most instances no clear evidence of maltreatment will emerge.

► Bed sharing is a common finding in many cases of SUDI. Epidemiological studies suggest that co-sleeping does increase the risk of SUDI in very young infants, if the parents smoke, or have consumed alcohol or sleep inducing drugs, and in other specific environmental circumstances.¹¹ Co-sleeping may affect the risk of death through effects on respiratory or temperature regulation, altered parental or infant arousal responses, or through direct risks of suffocation.

► In every case of sudden infant death, the parents will have their own needs for support, information, and grieving. Even where there are suspicions or clear signs of child abuse or neglect, the family deserves to be treated with respect. Most families express some feelings of guilt, and this may be even more prominent where there have been failures in care. Such parents need sensitive care to enable them to work through these difficulties regardless of our personal perceptions of their individual culpability. In this case, we acknowledged the concerns around Emma's substance abuse and previous neglect and the need for a full investigation of every aspect of Jordan's case, which effectively led to an even greater effort to support Emma and give her time and understanding along with feedback as information became available.

Table 3 Factors that may point towards maltreatment^{†*}

The American Academy of Pediatrics suggests that certain circumstances should indicate the possibility of intentional suffocation, including:

- ▶ previous recurrent cyanosis, apnoea, or ALTE while in the care of the same person
- ▶ age at death older than 6 months
- ▶ previous unexpected or unexplained deaths of one or more siblings
- ▶ simultaneous or nearly simultaneous death of twins
- ▶ previous death of infants under the care of the same unrelated person; or discovery of blood on the infant's nose or mouth in association with ALTEs

[†]It is important to recognise that none of these findings are diagnostic of maltreatment, and in most instances no clear evidence of maltreatment will emerge.

ALTE, apparent life threatening event.

The paediatrician discussed the results of the home visit with the pathologist before the postmortem examination, which was carried out the following day. This revealed a well nourished male infant with no signs of any external injury and a normal skeletal survey. Examination of the lungs showed scanty pleural petechial haemorrhages, but no focal parenchymal lesions. There was a small residual patent foramen ovale, but no other cardiovascular abnormalities. The gross pathology was otherwise normal. Histology later revealed some mild hepatic steatosis and lipid deposition in pulmonary macrophages. These initial findings were fed back to the mother in a subsequent home visit by the paediatrician with the health visitor. Emma was informed that at this stage, no specific cause of death had been found but that some further investigations were still awaited. The other professionals were reassured that there were no indications of any maltreatment and the focus of further management was therefore on continuing support for the mother.

The pathologist noted that, as with many necropsies in sudden unexpected infant deaths, there were no specific findings to account for Jordan's death. Petechial haemorrhages of pleura, thymus, and pericardium are common in SUDI and do not point to any particular cause. The residual patent foramen ovale would not have been of any clinical significance. Yet the finding of lipid deposition in the hepatocytes and pulmonary macrophages was unusual and he thought that it might indicate an underlying disorder of fatty acid metabolism. The initial cause of death reported to the coroner was SUDI: unexplained pending further investigation. The coroner agreed that at this stage the pathologist could inform Emma that no specific cause of death had yet been found, but that there were some, possibly insignificant, findings that required further investigation. In view of the previous concerns around neglect and maternal substance abuse, it was reassuring to note that there were no features pointing towards possible abuse or neglect.

COMMENT

- ▶ The postmortem examination is a vital element in the overall investigation of all cases of SUDI. As well as satisfying the coroner's need to rule out suspicious deaths, it can help to elucidate natural causes of death, to rule out others, to provide information and reassurance to the family, and to contribute to our overall understanding of SUDI. They should always be performed by a trained

Sudden unexpected death in infancy: key points

- ▶ Sudden infant death syndrome (SIDS) is the largest cause of infant death in the post-neonatal period. It accounts for at least 50% of cases of sudden unexpected death in infancy (SUDI), the remainder being due to a large number of medical and forensic causes
- ▶ The term SIDS should be reserved for the sudden death of an infant under 1 year of age which remains unexplained after a thorough case investigation, including performance of a complete necropsy, examination of the death scene, and review of the clinical history
- ▶ A number of metabolic disorders may result in SUDI, including disorders of fatty acid oxidation and mitochondrial respiratory chain disorders
- ▶ All SUDI should be thoroughly investigated according to a standard protocol involving a joint paediatric-police approach and incorporating a careful history, a detailed scene examination, a postmortem examination, and a final case review to pull together all relevant information
- ▶ All families who have suffered an unexpected death need to be treated with dignity and respect

paediatric pathologist according to a clear, evidence based protocol.¹²

- ▶ There has been considerable variation in the terminology used to describe SUDI where no cause has been found. The Kennedy report recommended that where, in the light of initial findings, the pathologist feels that there is no clear or sufficient cause of death, the cause of death should be given as "unexplained pending further investigation". This allows the coroner to open and adjourn an inquest and to release the body for burial.

Cultured fibroblasts were sent for metabolic investigations. Raised concentrations of acylcarnitine were found suggesting a disorder of fat oxidation. In order to investigate this further, the Guthrie screening card was sent for tandem mass spectroscopy. DNA analysis showed that the child was homozygous for the common mutation of LCHAD (long-chain 3 hydroxyacyl CoA dehydrogenase).

COMMENT

- ▶ With improved diagnostic methods, inborn errors of metabolism are increasingly being identified in association with sudden death in infancy. As a group, metabolic disorders may be responsible for a significant proportion of SUDI, although each individual disorder is in itself rare, and only very rarely do inborn errors cause sudden death in a previously healthy infant or child. Features in the history that may point to an underlying metabolic disorder include failure to thrive, developmental delay, seizures, vomiting, and diarrhoea. It is unusual for these disorders to cause problems in utero and most children are born at term with a good birth weight. Necropsy findings include hepatomegaly, cardiomegaly, and cerebral oedema, and fatty changes in the liver, kidneys, and muscle may be found. However, these changes are not specific to metabolic disorders and may be found in acute dehydration and severe infection, for example.
- ▶ Acylcarnitines are usually undertaken on a blood spot by tandem mass spectrometry, although this may be done on cultured skin fibroblasts. DNA can be extracted from either a blood spot or from fibroblasts. A blood spot onto a Guthrie card is an essential part of the investigation of every SUDI (table 1).

► LCHAD (long-chain 3 hydroxyacyl CoA dehydrogenase) deficiency is an autosomal recessive condition caused by a defect in the β oxidation cycle. LCHAD is a recognised cause of SUDI, but may also present with acute encephalopathy, hypoglycaemia in infancy, hypotonia, or cardiomyopathy.^{13 14} Women who are carriers of LCHAD may present with acute fatty liver of pregnancy. Acute compromise in infants with LCHAD may be precipitated by fasting or intercurrent illness. In these situations the infant is unable to metabolise body fat stores to provide energy and rapidly develops hypoketotic hypoglycaemia. The cause of death in these cases is likely to be related to abnormal metabolites having a toxic effect on the heart rather than on the hypoglycaemia alone, although typically one might expect a lower blood sugar in a young infant presenting with such severe illness. Definitive diagnosis is through enzyme assay using cultured fibroblasts, or confirmation of the recognised genetic mutations.

Following completion of all further investigations, a final case review meeting was held, three months after Jordan's death.³ The GP, midwife, paediatrician, social worker, and pathologist were all present. The findings from the full investigation including the postmortem examination and metabolic studies were reviewed, and the final cause of death was attributed to a disorder of mitochondrial β oxidation. Ongoing support for Emma had been provided by the GP and social worker. Emma had continued to live with her mother, who was helping to monitor and support her methadone programme. She was maintaining contact with her two older children and arrangements were underway to enable them to spend regular weekends with her at the grandmother's house. The paediatrician and GP made a home visit to discuss the outcome of the review with Emma and she was provided with a written account in plain English. She was also offered genetic counselling and support for any future pregnancies (care of the next infant).

FURTHER READING

► Byard RW, Krous HF. Sudden infant death syndrome: overview and update. *Pediatric and Developmental Pathology* 2003;**6**:112–27.

REFERENCES

- 1 Mackway-Jones K, Molyneux E, Phillips B, Wieteska S, eds. In: *Advanced paediatric life support. The practical approach*, 3rd ed. London: BMJ Books, 2001.
- 2 Royal College of Pathologists and the Royal College of Paediatrics and Child Health. *Sudden unexpected death in infancy. Report of a working group convened by the Royal College of Pathologists and the Royal College of Paediatrics and Child Health*. Chair: Baroness Helena Kennedy QC, September 2004, London: Royal College of Pathologists and the Royal College of Paediatrics and Child Health. www.rcpath.org.
- 3 al-Alousi LM, Anderson RA, Worster DM, et al. Factors influencing the precision of estimating the postmortem interval using the triple-exponential formulae (TEF). Part II. A study of the effect of body temperature at the moment of death on the postmortem brain, liver and rectal cooling in 117 forensic cases. *Forensic Science International* 2002;**125**:231–6.
- 4 Fleming PJ, Blair P, Sidebotham P, et al. Investigating sudden unexpected deaths in infancy and childhood and caring for bereaved families: an integrated multiagency approach. *BMJ* 2004;**328**:331–4.
- 5 Willinger M, James LS, Catz C. Defining the sudden infant death syndrome (SIDS): deliberations of an expert panel convened by the National Institute of Child Health and Human Development. *Pediatr Pathol* 1991;**11**:677–84.
- 6 Sidebotham P, Fleming P, Blair P. Sudden unexpected death in infancy. In: David T, ed. *Recent Advances in Paediatrics* 22. London: Royal Society of Medicine, 2005.
- 7 Office for National Statistics. *Deaths 2002: Childhood, infant and perinatal mortality: Live births, stillbirths and linked infant deaths by ONS cause groups and mother's country of birth*. London: Office for National Statistics, 2002, <http://www.statistics.gov.uk/STATBASE/>.
- 8 Harper RM, Kinney HC, Fleming PJ, et al. Sleep influences on homeostatic functions: implications for sudden infant death syndrome. *Respir Physiol* 2000;**119**:123–32.
- 9 Levene S, Bacon C. Sudden unexpected death and covert homicide. *Arch Dis Child* 2004;**89**:443–7.
- 10 American Academy of Pediatrics. Distinguishing sudden infant death syndrome from child abuse fatalities. *Pediatrics* 2001;**107**:437–41.
- 11 Blair PS, Fleming PJ, Smith LJ, et al, and the CESDI SUDI Research Group. Babies sleeping with parents: case-control study of factors influencing the risk of sudden infant death syndrome. *BMJ* 1999;**319**:1457–62.
- 12 Fleming PJ, Blair P, Bacon C, et al. *Sudden unexpected deaths in infancy. The CESDI SUDI Studies 1993–1996*. London: The Stationery Office, 2000.
- 13 Pons R, Roig M, Riudor E, et al. The clinical spectrum of long-chain 3-hydroxyacyl-CoA dehydrogenase deficiency. *Pediatr Neurol* 1996;**14**:236–43.
- 14 Duran M, Wanders RJ, de Jager JP, et al. 3-Hydroxydicarboxylic aciduria due to long-chain 3-hydroxyacyl-coenzyme A dehydrogenase deficiency associated with sudden neonatal death: protective effect of medium-chain triglyceride treatment. *Eur J Pediatr* 1991;**150**:190–5.

ep45

ARCHIVIST.....

Talking about infertility to young men with cystic fibrosis

Some 95–98% of men with cystic fibrosis (CF) have azoospermia because of aplasia of the vasa deferentia. Pregnancy may be achieved, however, in nearly two-thirds of couples in which the man has CF by use of microsurgical epididymal sperm aspiration and intracytoplasmic sperm injection. Several studies have suggested that adolescents and young men with CF need, and want, more information about their sexual and reproductive health. A report from Melbourne, Australia (S Sawyer and colleagues. *Thorax* 2005;**60**:326–30) has emphasised the point.

Questionnaires were sent to 125 eligible men (not receiving palliative care) who attended the regional adult CF clinic and 94 were completed and returned. The responders were aged 18–54 years (mean 30.5 years) and 58 were married or in a relationship. Sixteen had had a lung transplant. Only one respondent (aged 19 years) did not know about the effect of CF on male fertility but knowledge was often incomplete. Most had first heard about the infertility from CF clinic staff at an average age of about 16 years but they felt they would like to have been informed earlier. Seventeen men reported having been very upset when first told about fertility problems and those first told when older were more likely to be upset.

Forty-nine men had had semen analysis, all at age 24 years or older. Three-quarters of respondents thought that semen analysis should be offered at 17–18 years. As adolescents, some men had been uncertain about the difference between infertility and impotence and a substantial number had concluded that they would have no need for contraception. Sixty-two men said they would like more information about reproductive options. Seventeen men had a total of 27 children though only seven were biological fathers, one by natural conception and six by use of micro-epididymal sperm aspiration. Sixty-five of the 77 without children reported that they would like to become fathers at some time.

Reproductive options should be discussed with older teenage males in CF clinics and semen analysis should be offered.