CURRENT CONTROVERSY

Sperm donor suffers years later from inherited disease

D O E Gebhardt

Dutch press sympathetic to handling by hospital of discovery that a sperm donor was suffering from an inherited disease

On February 27th 2002, Dutch newspapers reported a serious mishap at a hospital in Den Bosch. An apparently healthy sperm donor during 1989 to 1995 informed the hospital in 1997 that he had recently been diagnosed as suffering from autosomal dominant cerebellar ataxia (ADCA) of which he was unaware when donating the sperm. The severely disabling neurological disease, which has a very low incidence, does not occur before puberty and may remain latent for many years. It has been established that 50% of the children from a parent with ADCA will also inherit the disease. The director of the hospital immediately informed the inspector of the Ministry of Health that 18 children had been conceived with the sperm in 13 women. The director did not tell the parents of the genetic disease of the sperm donor until three years later, because he first needed the advice of geneticists, ethicists, and lawyers. This mishap was waiting to happen. Now that it has taken place, it is of interest to review the reactions in the Dutch press. First of all, it is notable that none of the authors (including editors and science journalists) blamed the director, F Croonen, of the hospital in Den Bosch or its staff for the occurrence of this tragic event. They also did not reproach them for telling the parents what happened.

On the 27th of February 2002, The Volkskrant reported that this incurable disease, (ADCA), which was spread by the use of sperm from a donor, could not be detected by genetic tests. The first symptoms, which are caused by the gradual shrinking of the cerebellum, are a disturbance in speech and locomotion. In the article, the director is reported as saying: “After the diagnosis was confirmed, intensive consultations were held with a number of Dutch geneticists and ethicists. These consultations took some time, but we wanted to present the parents with sound information. It is of course bad news and you cannot offer them any consolation. We also had to inform their general practitioner. Medically speaking there was no reason to make haste to inform the parents. The children are all still young (between seven and 13 years) and the disease will not appear for some time”. Director Croonen believes that it was just bad luck. “Anyone who has children can pass on a genetic disease. What makes the calamity so unique is the fact that so many children (18) have been conceived. Another problem is that not all of the parents have told their child that his or her father is not the biological father.” In this article, the fertility expert and chairman of the sperm banks in Holland and Belgium, Dr P Janssens said: “Nowhere is donor sperm tested for genetic abnormalities. It is, however, tested for the occurrence of AIDS, hepatitis C, and some venereal diseases. The donor is also questioned about his health and whether hereditary diseases occur in his family. Sperm used for artificial insemination is, in fact, safer than your or my sperm.” In the same newspaper, one day later, Dr Janssens said that in general the sperm of one donor is used for the conception of 25 children. Once the new law on artificial insemination with donor sperm is approved by parliament, this number will, however, be reduced to 10. Also the name of the biological father will, under certain circumstances, become available to the child.

In The Volkskrant of the 28th of February 2002, two journalists write that a donor wished to contact the children who were conceived with his sperm, but heard that his file had been destroyed. This is because in Holland medical files need only be kept for 10 years. Croonen said that in his hospital the files of sperm donors were kept much longer.

In Trouw of 28th of February 2002, Croonen reported that most experts advised him to tell the recipients of the sperm what had happened. But some experts had reservations about this frankness, even if this meant that the child did not know that it could pass on the disorder on reaching maturity and conceiving children. Croonen also had difficulties in tracing some of the couples who had a child obtained with sperm of the ADCA donor. The privacy rules made it impossible to use the town records. Fortunately, the privacy regulations have changed recently and it is now possible for hospitals to obtain the addresses of anyone who has been treated there.

Croonen has no regrets that the parents were only informed three years after he had been told what had happened: “Thus we gave the parents three years of blissful ignorance. However, if a similar case should occur again, we would not wait so long.” The medical ethicist G de Wert, (Trouw 27th of February 2002) believes that once a test for diagnosing ADCA is available, the child and not the parents should determine if the test should be used. This is because the child also has the right, if so desired, not to know whether it has a genetic disease. So far only one ethicist (D Egberts, Algemeen Dagblad (1st of March 2002) has mentioned the medical principle of primum non nocere and raised the question of whether the whole procedure of artificial insemination should be reconsidered. The editor of Trouw (1st of March 2002) on the other hand concluded: “Sperm without a risk does not exist. Neither from a donor nor from a man who has a natural relationship. And even if in the distant future a safe technique is developed to determine genetic aberrations, it is doubtful if its use is desirable.”

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A case is described of a man who discovered years after donating sperm for artificial insemination, that he had a serious genetic disease, autosomal dominant cerebellar ataxia (ADCA). This manifests itself only years after puberty and occurs in 50% of the progeny. The sperm had been used for the conception of 18 children in 13 women. The parents were only told three years after the insemination. This long time interval was unavoidable since many of the parents had to be found and the hospital director wished first to discuss the matter with his legal advisors and other experts.

In future, in agreement with the new law only 10 inseminations will take place with sperm from one donor. This will reduce further the availability of donor sperm.
COMMENTARY

Gebhardt reports the tragic circumstances of a sperm donor who fathered 18 children in 13 women, who subsequently was diagnosed with autosomal dominant cerebellar ataxia (ADCA).1 The ADCAs are a genetically heterogeneous group of conditions that overall affect fewer than one in ten thousand people.2 Currently 16 locations on chromosomes have been found where genes reside that have mutations resulting in ADCAs.3 Testing is generally available for 5 (SCA 1, 2, 3, 6, and 7). Testing for the common CAG expansion (the type of mutation in these five genes that lead to ADCAs is the presence of three bases C, A, and G that repeat over and over, CAG CAG CAG etc) in these five genes will identify the genetic cause in about half of all families affected by ADCA.4 The offspring of the sperm donor are at 50% risk to inherit the mutation and therefore go on to develop the condition. Autosomal dominant cerebellar ataxia usually has its onset in adulthood and causes progressive unsteadiness (ataxia) with the sufferer generally becoming wheelchair-bound and ultimately dying from the condition around 10–30 years after the onset.5

Could this episode have been prevented? In all donor gamete programs, a thorough family history should be taken to detect the risk of such occurrences. In the case described, it is possible that the gentleman was unaware of his family history—for example, because he was adopted, or that his family history was apparently negative due to the genetic mechanism of anticipation. Anticipation is well described for conditions such as the ADCAs will not be forward as donors, should mean that it may well discourage people coming forward as donors, should mean that conditions such as the ADCAs will not be screened for, even if tests are available. Time will tell!

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