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## Sperm donor screening needs to be overhauled

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Writing in the 26 May 2009 edition of BioNews, the Donor Sibling Registry reported about the recent birth of a severely handicapped and profoundly retarded baby girl who had inherited an unbalanced translocation from New England Cryogenic Center sperm donor D-250. Now, the same problem has been disclosed as occurring at the London Women's Clinic. A donor was found to be a balanced translocation carrier only after his semen had been widely commercialised. A couple had to destroy 22 embryos created over the period of a year, at a cost of over 15 thousand pounds, and undoubtedly with considerable emotional investment. Eleven other women were also treated with the abnormal sperm, and are likely to also be suffering enormous turmoil. Whether they miscarried, are pregnant or were unsuccessful at conceiving, all of them will be asking tough questions about how this could happen. Weren't they told by the doctors they trusted, that donor sperm is rigorously screened and a safer option for getting pregnant?

These two incidents, occurring with a few months of each other, bring a spotlight on one of the oldest and most basic methods of donor screening - the karyotype. This method of staining and visually inspecting the chromosomes has been revolutionized by chromosome-specific fluorescent dyes. This allows much more powerful and precise visualisation of the chromosomes, to spot breaks, deletions, and the transposition of even small pieces. Since the frequency of reciprocal translocation is one in 500 people, it is critical that this \$400 test be performed on all prospective donors. As these experiences sadly confirm, the repercussions of such failure to screen can be very severe indeed. Using a donor with reciprocal translocations is potentially even more likely to produce sick children than donors with autosomal dominant disorders. The chances for a chromosomally abnormal child are thirty out of thirty-two (Scriven1998).

For an organisation like the DSR who counsels the casualties of these sperm bank irregularities, there is immense frustration that even despite the enormously high price tag of donor sperm, sperm banks

are still attempting to cut costs and boost profits by omitting screening. Since current screening standards are, despite sperm bank claims to be rigorous, in fact extremely minimal, the DSR wonders if private enterprises are the right parties to be conducting sperm bank operations. Is there a link between the need to squeeze out maximum profit for shareholders and woefully insufficient and lax screening practices?

In any event, the DSR is now campaigning to overhaul the current screening model and introduce a new regime which would significantly increase the genetic testing of sperm donors to include, extending CF, Tay Sachs and Glycogen Storage Diseases to all donors regardless of background, since offspring with those conditions have been born to non-high risk group donors and many African-Americans have Caucasian ancestry and so could carry the mutations. Testing all donors for Fragile X with a recommendation that any potential donor within the premutation range should be excluded. Screening for the mutations known to cause breast and ovarian cancers, haemochromatosis, melanoma, coeliac disease, polyposis and non-polyposis colorectal cancers and polycystic kidney disease. In addition, karyotyping should be mandatorily required.

Besides genetic testing, the DSR is also seeking to implement a novel method for the release of sperm vials, so as to prevent multiple affected offspring in case of an unrealised mutation. The method known as '5&2' was devised by the DSR's Chairman and Medical Director, Dr. Kirk Maxey. '5&2' prescribes that after thorough and complete medical qualification of naive donors, they are permitted no more than five pregnancies. Remaining sperm vials are then quarantined until the youngest in the cohort reaches two years of age and completes an extensive pediatric evaluation, along with the other four half-siblings. If all are healthy and without detectable abnormality, the donor's sperm is re-permitted and can now be used for twice the number of prior conception before a donor is retired. Such a system has the merit of preventing large numbers of children being conceived, before it can be established that the resulting offspring are healthy.

The '5&2' system should provide reassurance to the users of sperm that the sperm banks are monitoring offspring health. It requires active cooperation with recipients to agree to participate in the program by reporting back the infant and childhood check-ups and by agreeing not to conceive a second child from the same donor until after the first group of half-siblings have been cleared. However, if a social contract is agreed with the recipients then it is likely that high compliance will be achieved. Certainly by tracking births and health via the DSR most of the information, if not all, could be obtained that way. Both donors and recipients need to realize the value of posting and sharing their updated medical information on the DSR, as in a recent survey of 136 sperm donors the DSR found that 86 per cent had never been contacted by their sperm banks for medical updates.

The DSR wishes to stress that it regards its new genetic testing regime as a minimum program of testing and not an ultimate screening protocol. The objective in donor screening should be orientated towards full genome testing as soon as such a possibility becomes economically feasible and our understanding of genetic mutations makes such an endeavor worthwhile. Predictably in the future,

recipients will also be tested, so that mutually recessive donors and recipients aren't paired with each other.

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