UK centres are not following the Royal College of Pathologists’ recommendations for storage of Guthrie cards: a national policy is needed

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Cyclopia and sirenomelia in a liveborn infant

Recently, Chen et al published the case of a stillborn infant who presented with cebrophyaly, alobar holoprosencephaly, spina bifida, and sirenomelia. We would like to describe a liveborn infant with cyclopia and sirenomelia, identified by the Spanish Collaborative Study of Congenital Malformation (ECMC) in the total of 245 863 births, which appears that the case is the first one published with cyclopia and sirenomelia, bringing to three the total published cases with different degrees of holoprosencephaly and sirenomelia. 1 Cyclopia and sirenomelia are gross alterations of early blastogenesis. Thus, as Optiz postulated and we reported, 1 the combination of the two conditions, which is lethal, is a polytypic response of the alteration of the primary field, mostly affecting the midline. We have also observed that mothers of infants with blastogenic defects have a higher proportion of previous abortions that may mask the recurrence risk. However, we consider that in cases of holoprosencephaly (with different degrees of severity) and sirenomelia, in the absence of abnormalities of gestational age, the recurrence risk at birth should be very low.

We think that it is important to present this case in detail since, at present, it is the only one that has the two most severe forms of alteration of the cephalic and caudal part of the embryo (cyclopia and sirenomelia), and also because it was identified among a series of consecutive births, which allows us the estimation of the birth prevalence of this entity.

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1 Chen CP, Shih SL, Liu FF, Jan SW. Cebrophyaly, alobar holoprosencephaly, spina bi-

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It is a salutary experience to open the index of a history book and find oneself cited (albeit only once, and then only in parenthesis), if for no other reason than that it reinforces one's awareness of the passage of time. The events related in Errol Friedberg's book took place within living memory and the author deserves credit for eliciting for us the accounts of those who were responsible for them. The discoveries themselves are, of course, fascinating. While the need for living things to have means of dealing with the chemical and physical threats to their genomes is old hat to us, and the ways in which they achieve genomic stability and fidelity are not conceptually difficult today, this was not the case in the early days of DNA repair. It was first necessary to know the gene’s composition, its structure (established by Dan Brown and Lord Alexander Todd in 1952), and its conformation (as shown by Watson, Crick, and Wilkins in 1954). Indeed there is a subtext to the story in that the giants in the field of molecular genetics (as it came to be known) were in general little interested in genomic repair and tended to regard such work as second rate, largely because it was often done in national laboratories established and maintained for the purposes of supporting atomic energy and weapons research. Yet DNA repair has contributed more than its fair share of major discoveries, excision repair, mismatch repair, and the SOS coordinated inducible response being the most exciting with implications that extend to modern clinical medicine. Even the classic work of Avery, McLeod, and McCarty in 1944 showing that genes are made of DNA was antedated by Hollaender and Emmons in 1941, who showed clearly that the wavelength of ultraviolet light that was most effective in inducing changes in genes coincided with the peak of absorption by nucleic acid.

Various controversies are dealt with as fairly as appears possible. It is understandable that Friedberg has no wish to disturb the amicable relations that he has with his colleagues. One senses, however, that there could be another even more entertaining history written that gave more prominence to the paranoiae, suspicions, and personal animosities of the characters who showed them "nerts and all".

While the field up to, say, 1970 is covered well, after that the coverage is selective. There is, for example, no more than a paragraph about the repair of ionising radiation induced double strand DNA breaks, despite the fact that the field is of major importance, and the commonality of mechanism with immunoglobulin gene rearrangement foreseen since around 1980 has been spectacularly demonstrated in the last few years. Although there is a full and almost complete discussion of the inducible SOS system (lacking only an acknowledgement of the contribution of Peter Emmerson in Newcastle), there is no account of the discovery of the molecular mechanism by which mutations are made by the SOS gene products. Obviously, a second and updated edition will be needed before Friedberg himself retires.

One final thought in the reviewer’s mind concerns the conceptual nature of the really significant advances described in this book. They were frequently made by bright people thinking beyond the confines of their field, often beyond what they were being paid to do, going out of their way to interact with other bright people who were needed for mental cross fertilisation. The advances were more often than not unpredictable only with hindsight. Rarely would they have been the subject of a successful grant application in today’s world.

BRYN BRIDGES

NOTICE

Embryos, Genes, and Birth Defects

The Institute of Child Health and Great Ormond Street Hospital for Children NHS Trust will hold a Short Course on "Embryos, Genes, and Birth Defects" on 5-7 May 1998. Course organisers: Professor Peter Thorgood and Professor Robin Winter. Speakers will include Steve Brown, Andrew Copp, Dian Donnai, Patrizia Ferretti, Icuan Hughes, Robin Lovell-Badge, Willie Rardon, Peter Scambler, Cherryl Tickle, Veronica Van Heyningen. Themes to be covered: Research strategies, Mapping syndromes to genes, Use of dysmorphology databases, molecular genetic approaches, Transgenic technology, Teratology as an analytical tool, Developmental mechanisms, Analysis of birth defects of selected organ systems. Fee £175. For further information please contact: Courses and Conferences Office, Institute of Child Health, 30 Guilford Street, London WC1N 1EH, UK. Tel: 0171 829 8692, fax:0171 831 0488, e-mail: Courses@ich.ucl.ac.uk

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