Conference report

Journal of Medical Genetics 1989, **26**, 141–142

European Society for Human Genetics. Satellite meeting on meiotic microspreading

Participants (in alphabetical order): A C Chandley (Edinburgh), O Gabriel-Robez (Strasbourg), M Garcia (Barcelona), M A Hultén (Birmingham), R Johannisson (Lübeck), U Mittwoch (London), C Richler (Jerusalem), A Rosenmann (Jerusalem), Y Rumpler (Strasbourg), R M Speed (Edinburgh), J Wahrman (Jerusalem), B M Wallace (Birmingham).

This small workshop, held at Aberdare Hall, University of Wales, on 3 July 1988, was organised to discuss problems of common interest to those involved in the analysis of meiotic prophase chromosomes by microspreading.

All the participants present were experienced in mammalian microspreading and impromptu and informal discussion took place on the problems encountered in technique and in the interpretation of anomalous synaptonemal complex (SC) figures in spermatocytes and oocytes of man and several other species.

A starting point for discussion, raised by R Johannisson, was the problem encountered in trying to identify accurately the stages of zygotene, when lateral elements (LEs) are associating for synapsis, and diplotene, when elements are desynapsing before the first meiotic division. A general consensus of agreement was reached among the participants that the zygotene stage in man is characterised by pairing initiation from ends of arms, short paired lengths of SC appearing first at the telomeres and complete SC formation then following. The initial separation of LEs at diplotene, by contrast, tends to occur mainly interstitially along SCs.

The morphology of the XY pair came under scrutiny from R M Speed, one of his questions concerning the reason for the overdeposition of silver positive material along the X and Y axes in spermatocytes of patients with partially or completely arrested spermatogenesis. In addition, and perhaps as a consequence of overproduction of a proteinaceous product from the XY pair, was the question of why the XY pair in EM spreads was seen more commonly in association with NOR bearing D and G group acrocentrics at the pachytene stage among chromosomally normal subfertile and sterile men. This same question was later addressed by J Wahrman and colleagues, who strongly support the

original hypothesis of J Forejt (for the mouse) that germ cell failure in subfertile men carrying Robertsonian translocations could relate to increased levels of association between NORs of the translocated chromosomes and the sex chromosomes. The idea would be that some reactivation of the normally inactive X chromosome might occur at meiotic prophase in these males under the spreading influence from the translocated active autosomes, thus causing disturbance in spermatocyte metabolism. An alternative view was expressed by A Chandley, who suggested that XY-autosome associations in sterile structural heterozygotes might be a consequence rather than a cause of germ cell failure. Further investigations are needed to resolve this issue. The high frequency of involvement of acrocentric chromosomes in male sterilising reciprocal rearrangements also suggests NOR-sex chromosome association as a cause or consequence of germ cell failure (Y Rumpler, O Gabriel-Robez).

Agreement was reached on the question of whether recombination nodules can be seen in human microspread preparations, all present putting forward the same general opinion that these structures could not readily be seen either in human spermatocytes or oocytes by any of the techniques currently being used in their laboratories. Y Rumpler reported the ease with which recombination nodules could, by comparison, be seen and counted in spreads prepared from spermatocytes of lemurs. U Mittwoch presented data from the mouse showing that pairing anomalies of various kinds (triple associations, single LEs, non-homologous pairings, partially synapsed SCs) were to be found at a much higher frequency in oocytes than spermatocytes, whether from chromosomally normal or chromosomally abnormal subjects. The female germ line appeared quite 'error prone' in terms of synapsis compared with the male. R M Speed reported similar findings for the human, but M Garcia, who had studied spread preparations from male and female rats, stated that rarely had she seen synaptic anomalies in the oocytes from that particular rodent. No explanation can currently be offered for this difference between species.

Lengthy discussion took place about the possible

significance of the thickenings and enlargements observed in EM spreads along unpaired axes of human sex chromosomes and autosomes, and sometimes referred to as 'sprouts' or 'excrescences'. M A Hultén suggested that these enlargements arise at specific pairing sites along axes in a late but determined attempt to bring LEs together following initial pairing failure. Her belief therefore was that late synapsis could be achieved along such regions of chromosomes. A C Chandley and R M Speed, however, suggested that such cells may be lost altogether from the germ line by cell death or atresia, as unpaired axes showing excrescences are rarely encountered at the very late prophase stages (diplotene) in man. No suggestions as to the nature or composition of excrescences were forthcoming.

Matters concerning the technical procedures adopted in the various centres around Europe for microspreading of germ cells also occupied a considerable proportion of time. The virtues or disadvantages of spreading on sucrose, saline, or detergent, of staining with silver nitrate or phosphotungstic acid (PTA), and the effects of pH, water, and the type of plastic coating applied to slides were also discussed. Each laboratory had its own preference and, having found a technique which worked, was reluctant to change to any other.

Although there were suggestions for standardising the technical procedures and providing a system of standardised nomenclature for aid in the description of spread preparations (Y Rumpler), time was short and the meeting was forced to a conclusion without discussion on these points. It was agreed, however, that the Workshop had been highly successful, and a second meeting in another year's time might be considered. This, however, should extend over two to three days and workers involved in the spreading of gametocytes of plants, insects, and domestic species should also be invited to participate. This next Workshop might also perhaps incorporate practical demonstrations of spreading techniques.

All involved in the Cardiff spreading workshop agreed that the coming together of specialists in the field, although for such a brief time, had been a valuable and highly successful exercise, and one well worthy of repetition at a future date.

ANN C CHANDLEY* AND Y RUMPLER†

*MRC Human Genetics Unit,
Western General Hospital, Edinburgh EH4 2XU;
and †Laboratoire d'Embryologie, Hospices
Civils de Strasbourg, BP 426, 67091 Strasbourg,
France.