The X linked muscular dystrophies

This special issue of Journal of Medical Genetics comes at a time when Duchenne and other X linked muscular dystrophies are the subject of intense interest to workers in human genetics. The combination of new molecular genetic techniques with classical genetic linkage and family studies has produced rapid progress in a field which had previously lacked focus and direction. Not only are the genes for all three main X linked muscular dystrophies now accurately localised, but in the case of Duchenne and Becker dystrophies we are beginning to understand the disorders at the molecular level, as well as having accurate, though not yet perfect, methods of carrier and prenatal detection.

The idea of a special issue was conceived after a meeting in London earlier this year, organised by the Muscular Dystrophy Group of Great Britain and described in the Conference report by Dr Kay Davies (p 482). Workers participating in the meeting responded enthusiastically to the suggestion and their contributions have been joined by others during the following months. It was decided from the beginning that the issue should consist principally of original work rather than reviews, and that contributors should submit work they were currently performing rather than attempt systematic coverage of the entire field. The result is probably a fair reflection of the range of genetic work in progress today, though inevitably biased towards that in the United Kingdom and towards molecular biology.

The importance of cytogenetic studies, both in the initial localisation of the Duchenne gene and in its subsequent analysis, can be seen in the first group of papers, while the interest and practical importance of molecular deletions detected by the pERT and XJ clones is reflected in the papers on this topic. The p21 region of the X chromosome must now be one of the most intensively mapped parts of any chromosome, and the linkage studies in this issue contribute still further to this, while the analysis of population dynamics emphasises the importance of relating new approaches to the foundations of classical genetics. The succeeding papers on prenatal diagnosis and carrier detection emphasise the real practical results that have arisen from our improved knowledge. Finally, the group of papers on Emery-Dreifuss muscular dystrophy show that the heterogeneity which disappeared with the demonstration of a similar localisation for Becker and Duchenne dystrophies is now a reality again, with a distinct locus at q27–28 for the Emery-Dreifuss form.

At the time of writing, the specific genes and gene products for the X linked dystrophies are still unidentified*, and the probable complexity and instability of the p21 region provides a major challenge in achieving their identification. Nevertheless, the intensity of research reflected in the papers presented here gives reason for real optimism, not just for our understanding, prevention, and ultimate treatment of the X linked muscular dystrophies, but also for similar progress in the many other genetic disorders which are now approachable at the level of the gene itself. Similarly, the high degree of collaboration that has evolved between different groups, as well as between basic scientists and clinicians within these groups, should serve as an excellent example for workers concerned with other disorders.

*As this issue goes to press, it is clear that the work of Kunkel and colleagues, presented at meetings and in press in Nature, has identified at least part of the gene itself.

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