Phase transitions

Cosmology in the laboratory

from T. W. B. Kibble

AT FIRST sight it may be surprising that useful information about the processes leading to the formation of the largest structures in the Universe could come from small-scale laboratory experiments. Yet just such a bold proposal is made by W. H. Zurek on page 505 of this issue[1].

The proposed experiment is a very simple one. Apply a rapid pressure quench to a sample of liquid helium in an annular container to take it quickly through the transition from normal to superfluid. Zurek predicts that the sample will acquire a random non-zero circulation velocity which should be readily measureable. If confirmed, this will be of considerable interest in its own right in terms of the physics of superfluids, but the motivation for the proposal comes from the need to test our ideas about cosmic strings — a remarkable example of fruitful fertilization between very diverse disciplines.

Phase transitions occurring in the very early stages of the history of the Universe (only a fraction of a second after its birth) may generate various topologically stable defects[2] such as monopoles, domain walls and in particular strings, which are analogous to the quantized vortex filaments in superfluid helium or the flux tubes in superconductors. It was suggested by Zel'dovich that these cosmic strings might provide the initial density perturbations that lead to gravitational condensation of galaxies. This idea was refined by Vilenkin[4,5] and others into a very attractive theory of galaxy formation. Recent, as yet unpublished, work by Turok has shown that it can explain rather well the correlations of large clusters. The theory depends, however, on an assumption about the density of strings produced at the phase transitions[6,7], which is of course quite inaccessible to direct observations. Zurek's idea is to test the assumption on superfluid helium.

When liquid helium undergoes the transition from normal to superfluid, the complex order parameter ψ, which can be thought of as the wave function of the Bose condensate, acquires a non-zero value. If the transition is sufficiently rapid, the phase of ψ will vary randomly from one region to another and vortex lines will be produced. These are trapped regions around which the phase varies by a multiple of 2π. Since the superfluid velocity is proportional to the gradient of the phase, the lines carry quantized vorticity.

The vortex lines once produced will evolve rapidly and leave no direct evidence of their initial distribution. But Zurek points out that if an annular geometry is chosen, there may be a non-zero net circulation around the annulus. Because vorticity is quantized, this flow will persist and may lead to a circulation velocity in the superfluid of the order of millimetres per second, which should be readily measurable.

Even if this prediction is confirmed in relation to superfluid helium, why should we believe that it tells us anything about cosmic strings? After all, the transition temperatures differ by factor of perhaps 10^{28}. The answer lies in the universality of behaviour near a phase transition. Liquid helium is well described qualitatively by the Landau–Ginsberg model. The relativistic version of this is the Higgs model, the paradigm for discussions of spontaneous symmetry breaking in field theories. Provided the symmetries of the problem are the same, we expect the same qualitative behaviour, despite the very different scales. Vortex lines should be a good analogue of at least one class of strings.

With regard to the most popular type of cosmic string, however, there are important differences. Most significant is the fact that the symmetry involved in the superfluid case is a global one, not a local gauge symmetry. In particular, there is no gauge field coupled to the relevant current. In this respect, a better analogue would be a flux tube in a superconductor. It would be fascinating if one could perform the Zurek experiment in a superconducting ring, but it might be very hard to induce a sufficiently rapid transition. In helium, the existence of another thermodynamic variable,the pressure, is crucial. Magnetic field could not be used in a similar way, because the goal would be to generate magnetic field spontaneously.

Particle physicists and cosmologists will eagerly await the results of the Zurek experiment. The expected positive result would be a considerable boost for the string theory of galaxy formation. □

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Human genes

Mapping hereditary disorders

from Albert de la Chapelle

THE rate of progress with which human genes are being allocated to identified regions of individual chromosomes is rapid. In four years, the number of mapped genes has increased nearly three-fold to over 900. Sixteen mapped genes had been cloned four years ago; the present figure is 249. And the number of regionally localized DNA segments ('probes') that are not known to represent genes but are an important aid to mapping has risen from some 50 to over 800. These figures were the raw facts of a recent workshop* that illustrated how knowledge of the human gene map helps to clarify the molecular basis and pathogenesis of many mendelian disorders. Moreover, with the aid of the gene map, disorders can be diagnosed before they are molecularly understood.

Cystic fibrosis (CF), one of the most common recessively inherited mendelian disorders of Western populations, has an incidence at birth of approximately 1 in 2000 (with a remarkable gene carrier frequency of about 1 in 20). Nevertheless, ignorance about the *CF* gene has been virtually total. The first lead, reported at Helsinki, is evidence of a linkage between the *CF* gene and the *PON* gene for the enzyme paroxonase [1]. The measure of the reliability of the linkage, the 'lod score', is 3.38, where lod scores in excess of 3.0 are considered 'formal proof' of linkage. The data suggest that the genes for *CF* and *PON* are located approximately 10 recombination units apart on the same, so far unidentified, chromosome. Once the *PON* gene has been cloned, it should be possible to determine the chromosome and band where it and *CF* are located by the methods of classical somatic-cell genetics or *in situ* hybridization. Then chromosome-specific libraries will be screened for closely linked probes, which should lead more quickly to the *CF* gene than chromosome 'walking' or 'hopping' from *PON* to *CF*, since the genetic distance of 10 recombination units is long, corresponding to some 10^7 base pairs.

If the *CF–PON* linkage is confirmed, it will certainly represent a breakthrough in CF research but, unfortunately, the PON phenotypic polymorphism is quantitative rather than qualitative. It remains to be seen how much overlap there is in the observed biomodal distribution of paroxonase concentrations in the plasma and whether it is really due to alleles at the structural *PON* locus.

Another inherited disease of unclear aetiology and pathogenesis is adult polycystic kidney disease (PKD1).Its gene frequency is approximately 1 in 1000 and, as

*The Eighth International Human Gene Mapping Workshop, Helsinki, Finland, 4–10 August, 1985. Proceedings will be published as vol. 40 of *Cytogen. Cell Gen.* this year.