NEWS

These included diffusion-limited aggregation model (DLA), ballistic-driven aggregate growth (BDAG), etc. He gave many examples of growth of real 'fractal' objects. In the last invited talk on magnetic studies using heavy ion beams, S. N. Mishra showed a novel use of accelerator beams for probing magnetic ordering in a host material through the use of isomeric nuclei produced by a nuclear reaction and then looking at time differential angular distribution of gamma rays or TDPAC. He brought out the strengths and limitations of the method and presented a host of data on the formation of local moments in a number of systems.

The second session was devoted to oral presentations with a fairly wide coverage of different topics such as density functional calculations, X-ray scattering, neutron scattering, models for crystallization, anisotropic X–Y model, etc. The organizers had to accommodate a large number of poster papers and therefore the time available for discussion was restricted. To somewhat mitigate this difficulty poster sessions were so arranged that papers on the same topic were distributed over different days. Some of the broad features which emerged from the sessions may be recorded as follows. About one third papers were not presented as the authors were absent. The experimental papers outnumbered theoretical ones by almost five to one, showing that the experimental facilities for condensed matter research have greatly improved in the last decade. A number of studies on phase transitions were reported using NMR, EPR, TDPAC techniques. A variety of film deposition techniques like MBE, RF sputtering, LB method, etc. were covered. Magnetic studies in intermetallic alloys, spinels, etc. were reported using magnetometry, Mössbauer spectroscopy, neutron diffraction, etc. Studies in many liquids and liquid crystals were reported using NMR, Raman, XRD and dielectric constant measurements, etc. Amongst $\text{Hi–T}_2$, materials study of irreversibility line, looking for dimensionality effects and role of synthesis conditions on the properties of these materials were reported by several groups. Persons interested in details could consult the book containing extended abstracts of the papers and printed under the title 'Proceedings of the DAE Solid State Physics Symposium', Volume 36C. The invited talks are going to be published as a Special Issue of the Indian Journal of Pure & Applied Physics published by the Publication Directorate of CSIR, New Delhi.

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RESEARCH NEWS

Genetic defects in hereditary motor sensory neuropathies

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Neuropathies consist of a group of diseases of peripheral nerves resulting in weakness and wasting of muscles of the limbs, impairment of various modalities of sensations or autonomic functions either alone or in combination. The causes of neuropathies are diverse. The loss of function of nerves results from the degeneration of axons of nerve fibres (axonopathies) or loss of myelin, the material around nerve fibres produced by specialized cells called Schwann cells (demyelinating neuropathies).

Electrophysiological tests can detect abnormalities of nerve function such as slowing of nerve conduction velocities which is seen in demyelinating neuropathies or loss of amplitude of action potentials generated in the muscles in response to electrical nerve stimulation, in axonopathies.

Nerve biopsies in demyelinating neuropathies reveal segmental loss of myelin of nerve fibres and reduction in the number of myelinated fibres. Chronic demyelination and remyelination can result in the formation of 'onion-bulb' appearance due to ensheathing of single nerve fibres in several layers of Schwann cell processes.

The hereditary motor sensory neuropathies (HMSN) are a heterogeneous group of inherited neuropathies whose etiology was hitherto unknown. Rapid advances in molecular biological techniques in the last few years have improved the understanding of the genetic defects in some types of HMSNs.

The major types of HMSNs are types I, II and III. HMSN type I presents in the first or second decade of life. Genetically two distinct forms, autosomal dominant (AD) and autosomal recessive (AR) have been well identified. In some families of HMSN I, the disease has been linked to the Duffy blood group on the long arm of chromosome 1 and have been designated as HMSN Ib. They constitute only a minority of patients with HMSN. Most families of HMSN I are not linked to the Duffy locus (HMSN type Ia) and have the disease locus on chromosome 17. There are also reports of a non Ia–non Ib form, in which loci on chromosome I and 17 have been excluded.

Molecular biological studies in Trembler and allelic Trembler J mice, the animal models of demyelinating neuropathy, have thrown light on the pathogenesis of AD type of HMSN Ia. These mice have features of an autosomal dominant demyelinating or hypomyelinating neuropathy with localization to a region in chromosome 11 of the mouse genome, which is homologous to human chromosome 17.

In these mice, two-point mutations have been found in the newly described myelin gene, the PMP-22 gene, the expression of which is demonstrated only in the peripheral nervous system of mice. The point mutations comprise of substitution of aspartic acid for glycine and proline.
HMSN type I resembles the AD form but the genetic defect or defects resulting in the disease have so far not been discovered.

HMSN type II (neuronal form) is also inherited in an AD or AR pattern. It clinically resembles type I disease to a large extent but is less commonly encountered. Pathologically it is an axonopathy and recent detailed studies have shown loss of motor neurons in the lower spinal cord and sensory neurons in the sensory ganglia resulting in secondary degeneration of nerve fibres originating from these neurons. The genetic defect and metabolic abnormalities responsible for the neuronal loss are yet to be identified. The proximal 1q site of HMSN type II and 17p. 11.2 site of type Ia have both been excluded.

HMSN type III is a rare and more severe form of demyelinating neuropathy presenting in early childhood.

Classical onion-bulb appearance is seen in nerve biopsies and nerve conduction velocities are severely slowed. Recently two other sub types of HMSN type III with AR mode of inheritance associated with absence of myelination of nerve fibres in one and basal lamina onion bulbs in the other have been described.

Though the possibility of sex-linked inheritance of HMSN has been suspected for a long time it was confirmed only recently. Linkage analysis conducted in several kindreds suggests that the gene for X-linked dominant HMSN is located in the X-chromosome on the proximal long arm close to the centromere in between the phosphoglycerate kinase pseudogene (Xq 12) and the DXS 72 locus which maps to Xq 21.1.

Being an X-linked dominant disease, male-to-male transmission is never seen while the affected females transmit the disease to half of their offsprings of either sex. Clinically it resembles AD HMSN type Ia but males are more severely affected than females.

There are also reports of an X-linked recessive form of HMSN but evidence for linkage to X-chromosome is not definite.

Hereditary neuropathy to pressure palsies (HNPP) is an AD disorder characterized by recurrent focal neuropathy triggered by minor nerve trauma, which begins in adolescence. Nerve conduction studies reveal conduction block or slowing in the affected as well as unaffected nerves. Pathologically there is sausage like thickening of myelin sheaths and segmental demyelination.

The HNPP locus has also been assigned by linkage studies to band 17 p. 11.2 as in HMSN Ia. However, in HNPP, this region is deleted and patients are monosomic for this segment on chromosome 17. This was seen in affected members of 3 families and de-novo deletion documented in one pedigree.

This observation further supports the hypothesis that the alteration in the PMP-22 gene located in this segment is responsible for the phenotypes of HMSN Ia and HNPP. The exact role of the PMP-22 gene on axon-myelin interaction and the sequence of events that lead to the breakdown of functionality and morphologically intact myelin in these two conditions have to be better understood. Similarly the occurrence of genes other than PMP-22 in the region 17 p. 11.2 and mutations in other peripheral myelin proteins may have to be looked for in order to understand the pathogenesis of other types of HSNs.


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