Genetics

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Genetics touches every aspect of biology – the boundaries between genetics, molecular biology and biochemistry have become diffuse and their application and exploitation in viral, microbial, animal, plant and environmental science provides the scientific knowledge base for current and future research at SCRI. The new Genetics Division brings together expertise in plant breeding, genetics of crop plants, molecular genetics, genomics and gene expression. Interactions, both within the Division, and with chemists, biochemists, pathologists, cell biologists and environmental scientists in other Divisions, is the key to producing high quality research in the future, which is competitive nationally and internationally.

The success of breeding programmes has been based on an understanding of the biology and genetics of particular plant species, their interactions with pathogens, the ability to phenotype traits, plant handling and field trialling skills, and quantitative genetic and statistical analyses. Capabilities and expertise in gene cloning, gene expression and plant transformation, and the development of germplasm and molecular markers, built up over the last 10 years at SCRI, provide the basis for establishing structural and functional genomics research. This will greatly impact our understanding of the developmental and evolutionary biology of plants underpinning traits of commercial importance. As the sequences of most plant genes are identified through genome sequencing of *Arabidopsis* and rice, and high throughput genomics approaches to gene discovery and function, new opportunities arise in both fundamental and applied genetic research. The new genetics will ultimately allow us to dissect the molecular and biochemical events which give rise to complex phenotypes of economic importance, and identify genes involved in these processes. In the future, more emphasis will be needed on genetic and biochemical approaches to address gene function of isolated genes for such exploitation. In the meantime, the development and extensive application of marker systems will provide ever-increasing resolution of genome organisation, more precise mapping of molecular markers, and the potential to assess variation in wide germplasm. SCRI maintains large germplasm collections of potato, blackcurrant, raspberry, and, to a lesser extent, barley. One of the future goals of the Genetics Division is to characterise fully the germplasm genetically and phenotypically to provide information for breeding objectives in plant improvement programmes in the short, medium and long term.

At present, molecular markers are being utilised to develop and assess marker-assisted selection of parents and progeny in barley. The application of markers to established, accelerated, breeding methods in potato will give a greater understanding of the mechanisms behind the success of these methods, providing the potential for further improvement. Taken together

with identification of genes involved in major traits, the assessment of genetic and functional variation of alleles in diverse germplasm and gene-specific markers, the potential for targeted and rapid plant improvement based on extensive parent and progeny marker-assisted selection will continue to increase. The efficiency of the breeding process will aid in overcoming some of the limitations of traditional plant breeding, such as breeding barriers and unfavourable gene linkages, and provide incredible opportunities for targeted plant improvement not feasible by traditional approaches.

The vast quantities of data being assembled locally and internationally require us to build a Bioinformatics infrastructure and capacity. Data on markers, genotypes, genetic maps, DNA sequences and expression profiles are being generated locally and need to be assembled in a highly usable and accessible format. Local databases must be compatible with other national and international databases. Scientists at SCRI must have access to information on all aspects of relevant biology to inform research programmes and their direction. There is no doubt that genomics and bioinformatics will change the way in which scientific problems in the future are addressed.

To address gene function, a battery of techniques are being or have been established. A key technology is plant transformation where over-expression or antisense knock-out are used to assess function. The ability to transform with isolated genes also provides a second route to plant improvement, overcoming breeding or crossing barriers and allowing introgression of specific genes. This depends on our ability to transform the mandate crops and to isolate genes and understand mechanisms of gene expression and its regulation.

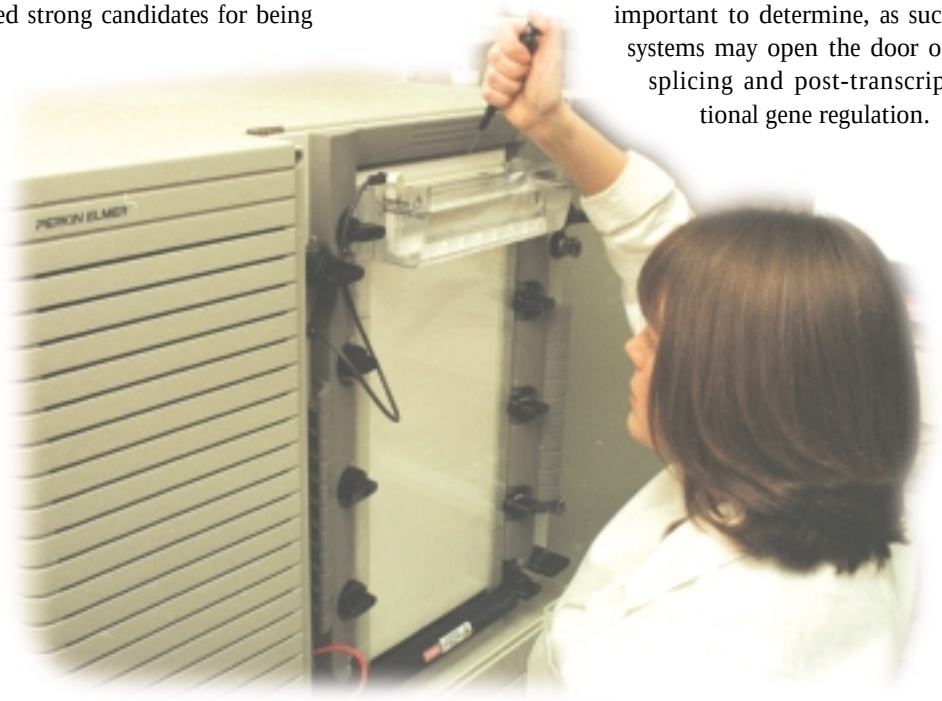
The expansion in structural genomics must also be paralleled by increased molecular, genetic and biochemical analysis of key biological processes and by increased capabilities in gene function technologies such as virus-induced gene silencing and RNAi (Biochemistry/Cell Biology and Pathology Divisions). Plant genomics will be paralleled by research on pathogens and pests, and plant-pathogen interactions in the Pathology Division. The technologies to analyse relationships between cultivars and related species, and to identify novel sources of genetic diversity, can also be applied in studies assessing the effects of environment on genetic diversity within plant systems (Plant, Soils and Environment Division). The increased effort will require the establishment and management of interactions between innovative scientists. The establishment of generic genomics tools and technologies using mandate crops, in particular barley and potato, provides SCRI with flexibility in the future to apply the technology to other plant or crop species in response to changing patterns in agriculture.

Against a background of reduced profitability in agriculture, plants will remain a source of new products for processing and food industries. The diversification of plants to produce novel compounds for downstream industries will depend on innovative ideas from teams of chemists, biochemists, molecular biologists, geneticists and breeders with knowledge and expertise in metabolic and biochemical pathways, gene systems, gene expression and introgression of genes and traits into advanced and adapted germplasm. The challenge for the future is to maintain the knowledge of specific plant/crop systems, the ability to assess and measure phenotypic variation accurately, and breeding expertise, and marry these to the exponentially increasing knowledge and capabilities of the new genetic and genomic approaches.

Genomics Unit A major objective of plant genomics is to discover all of the genes in an organism and determine their location on genetic and physical maps. By correlating the location of genes with the location of loci affecting traits, it is possible, after a number of parallel studies, to establish the role of a gene in a given process (the candidate gene approach for gene identification). Our studies focus on both potatoes and barley. Using potato as an example, over the last year we have made four significant advances towards these objectives. First, using a combination of AFLP and SSR markers and a diploid segregating population, we have constructed an ultra-high density genetic map (UHD map) of the potato genome comprising over 6,000 mapped markers (in collaboration with colleagues in The Netherlands, France and Spain). The target is 10,000 markers, which represents approximately 10 markers per centiMorgan (cM is the unit of genetic distance). Second, we have constructed bacterial artificial chromosome libraries of one of the parental clones (BAC libraries contain large size DNA inserts and are a key component of physical mapping strategies) and have developed a pooling strategy which allows us to identify groups of BAC clones which support the amplification of the 6,000 AFLPs already placed on the genetic map. Thus, in principle, we can quickly build thousands of short physical contigs (a contig is a group of physically over-

lapping large DNA clones) and link them directly to the genetic map. Currently, we are using this strategy to build a physical map of a region of the potato genome covering approximately 2-5 cM of the genetic map, which is known from previous studies at SCRI and elsewhere to contain a number of genes influencing a wide range of phenotypic characters such as dormancy, yield and disease resistance. Once the physical contig is built, we will sample sequence the entire region to identify all of the genes and, if funding allows, proceed with complete genomic DNA sequencing of this region. Third, we have completed a pilot expressed sequence tag (EST) project which has generated the partial DNA sequence of approximately 2,500 potato genes. By exploiting the physically mapped BAC clones, we will be able rapidly to position the majority of the ESTs directly on to the physical and genetic maps. From the alignment of these maps, we will ultimately be able to relate the position of genes (and identified but uncharacterised contigs containing genes) with the position of genetically mapped phenotypic traits. Genes which are functionally consistent with having a role in the trait, are expressed at the relevant time and, in the relevant tissues, can be considered strong candidates for being components of the trait. Candidate genes will then have to undergo a range of other studies involving plant transformation. Finally, the volume and complexity of this type of data demands that sophisticated methods for acquisition, archiving, analysis and interpretation of biological information be developed and implemented. Over the last year, significant progress has been made towards the automated processing of molecular markers, DNA sequence and BAC fingerprint data. Approaches for integrating such diverse data types and archiving these in an accessible and informative manner are currently being explored.

Gene Expression Unit Regulation of gene expression at the post-transcriptional level is complex and subtle. A major achievement of the RNA Processing Group over the last 2 years is the characterisation of a plant mini-exon system. In contrast to vertebrate mini-exon systems, inclusion of the potato invertase mini-exon is regulated by strong plant intron splicing signals instead of specific regulatory elements. This splicing system is very sensitive to sequence changes in the splicing signals, and provides the best system to date for systematic characterisation of plant splicing signals, which has previously been very difficult. In addition, proteins which bind to the invertase transcripts *in vitro* are being examined. In parallel, in collaboration with scientists in Switzerland and Austria, who have isolated genes for a number of protein splicing factors, proteins which alter splicing behaviour *in vivo* can be characterised. Thus, the potato invertase mini-exon system is proving extremely useful in addressing outstanding questions on plant pre-mRNA splicing. A number of other mini-exons (as small as 3 nt) have been described in plants. Whether such transcripts depend on similar mechanisms or involve novel regulatory elements and factors is important to determine, as such systems may open the door on splicing and post-transcriptional gene regulation.



The continuing focus on gene expression during the malting process of the barley grain has led to developments in several areas. A foundation unigene set derived from 2000 expressed sequence tags (ESTs) forms the basis for initial microarray studies on gene

expression during the malting process. Genetic diversity is being determined for a subset of these unigenes by their amplification and sequencing across a variety of barley germplasm, from mapping parents to landraces and wild barley. During this exercise, the prevalence of single nucleotide polymorphisms will be established and opportunities explored for their practical utilisation. Key ESTs have generated new research programmes, both at SCRI and elsewhere, on individual genes of interest both from pure and applied science viewpoints. New mapping and transgenic technologies are also being developed for the exploitation of this gene sequence information. This research aims to develop the interface between traditional hypothesis-driven experimental molecular biology and high-throughput non-biased data collection strategies central to genomics efforts as described above. It is anticipated that this will be fertile ground for practical crop improvement programmes.

Applied Genetics Unit Research in the Applied Genetics Unit is focused on SCRI's mandate crops, comprising barley, potato, raspberry and blackcurrant. Contract research and breeding is also carried out on brassicas (oilseed, forage and vegetable), legumes (*Faba* bean) and strawberry. The aim is to use modern molecular genetics and genomics to understand and exploit the genetical variation available in crop species and their wild relatives, and to study the genetics of economically important traits in order to underpin scientific crop improvement for yield, quality and resistance to diseases, pests and abiotic stresses. SCRI maintains the Commonwealth Potato Collection and *Rubus* and *Ribes* germplasm collections, and has access to wild barley germplasm. Malting quality in barley, processing quality in potatoes, and berry quality in soft fruit are all currently important targets. As most of these economically important traits display continuous variation, the emphasis is on developing the theory and practice of quantitative trait locus (QTL) analysis and of molecular marker-assisted selection (MAS). The Unit has developed and mapped doubled haploid populations of barley and F1 and backcross populations of potatoes at the diploid and tetraploid level, and is developing F1 populations of blackcurrant and raspberry. A diagnostic marker has already been developed for BYMV resistance in barley and is being used widely in commercial breeding programmes in Europe. The research leads to improved parental material for use in commercially-funded breeding programmes, and also to faster, more efficient and novel components of breeding programmes which can be tailored to meet the requirements of commercial customers. It is through these commercial contracts that we are able to produce finished cultivars that meet the ever increasing demands of farmers, growers, processors and supermarkets, as well as addressing public concerns about healthy food and a safe environment.