transplant related mortality (TRM) of 9% overall was observed, with inter disease differences (12.5% in SSc and only one TRM in RA). Around 2/3 of patients responded with improvement or stabilisation. In SSc, 69% achieved a 25% or greater improvement in skin score, and in secondary progressive MS, 78% a 3 year progression free survival. Relapse was more common with RA and JIA, mostly being a synovitis easy to control with agents which were ineffective pre transplant. Randomised, controlled prospective trials are underway in SSc (ASTIS Trial: HSCT versus monthly pulse cyclophosphamide)), and planned for RA and MS. Trials in JIA are being discussed and further phase I/II data are required for SLE and vasculitis. These will be presented, together with immune reconstitution data, sometimes predicting relapse, and new developments in HSCT, especially non myeloablative "mini" allograft techniques. This may be applicable to full sibling matched selected patients.

REFERENCE

- 1 Biggs M, Passweg J, Furst D, *et al.* Phase I trial of autologous stem cell transplantation in systemic sclerosis: procedure related mortality and impact on skin disease. *Ann Rheum Dis.* 2001, in press

Ankylosing Spondylitis – Thursday 14 June, 14.00-15.30/Congress Hall

SP0031 GENETIC STUDIES IN FAMILIAL ANKYLOSING SPONDYLITIS: THE NORTH AMERICAN SPONDYLITIS CONSORTIUM EXPERIENCE

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Recent genome-wide screens of English families multiplex for ankylosing spondylitis (AS) have suggested genes additional to HLA-B27 to be involved in disease susceptibility, particularly on chromosome 2q and 16q. Another recent study has suggested an association of AS with an allele of the IL-1 receptor antagonist gene (IL-1RA) on chromosome 2q. In order to confirm these findings in an independent group of families, 92 sib pairs concordant for AS by modified New York criteria from 74 North American families from the were examined with eight polymorphic microsatellite markers within the major histocompatibility complex (MHC) and HLA-DRB1, DQA1, DQB1 alleles; four markers on chromosome 2q and three on 16q near the regions previously implicated. Results for the MHC markers were as follows:

Abstract SP0031 Table 1

Marker	Map position (cM) ^a	NPL Statistic	p-value
D6S461	40.14	2.6	0.005

D6S276	44.41	3.09	0.001
D6S105	44.41	2.17	0.015
D6S273	44.96	3.17	0.001
D6S1014	45.5	1.56	0.023
D6S1666	45.5	3.03	0.001
D6S1583	47.71	5.38	8×10^{-6}
DRB1	49.87	3.17	9×10^{-7}
D6S1019	53.81	2.09	0.018

No evidence of linkage was found for the markers on chromosome 2q and 16q. In addition, analysis of 29 single nucleotide polymorphisms (SNP's) from the IL-1 RA gene in 30 AS patients and 48 controls found no evidence of disease association. These data strongly suggest the presence of a second MHC gene operative in predisposition to AS at or near the HLA-DRB1 locus. Additional families are currently being studied and fine mapping studies in progress to confirm and extend these findings.

SP0033 UPDATE ON PSORIASIS AS IT RELATES TO SPONDYLOARTHROPATHIES

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Psoriasis is a common disease (affects ~ 2% whites); and up to 10% of these patients have associated inflammatory arthritis [psoriatic arthritis (PsA), including spondyloarthropathy (SpA)], which may, in up to 15% of patients, precede the onset or diagnosis of psoriasis. There are no internationally agreed criteria for the diagnosis of (PsA); it has been broadly defined by Moll and Wright as an "inflammatory arthritis associated with psoriasis, which is usually negative for rheumatoid factor". More precise criteria and a large case-control study comparing patients with PsA with controls without arthritis (despite having had psoriasis for >10 years), properly matched for age, race, gender, geographical area, and for the subtypes of psoriasis/arthritis are needed. It would be advisable to have another control group with inflammatory arthritis without psoriasis.

The sibling recurrence risk (lambda s value) is estimated to be roughly 4 for psoriasis. Self-reported positive family histories for psoriasis among PsA patients have been observed in up to 40% and for PsA in 10%, but the familial aggregation does not follow a simple Mendelian trait. Psoriasis and PsA have not only clinical but also genetic heterogeneity, and multiple susceptibility alleles at different loci may produce the same phenotype, resulting in clinical misclassification of the disease type. There are, as yet unknown, genetic and environmental interactions, and also different combinations of susceptibility genes may predispose to different patterns of psoriasis and PsA (variable disease expression), and not everyone with such genes will develop the disease (variable penetrance). Psoriasis is associated with HLA allele Cw*0602, MICA allele 5.1, corneodesmosin allele 5 (CD*5), OTF3 beta, and HCR (pg8); they are all located on chromosome at the 6p21 region. Associations with chromosomes 17q25, 4q32-35 and 1q (S100A7) have also been reported. Similar studies of affected sib-pair families with PsA are also needed.

Activated T lymphocytes and dendritic cells mediate the disease, but the antigen (s) responsible are not known. Although