Abstract – Primary Lymphoedema

Primary lymphoedema will arise from a genetically determined abnormality of the lymphatic system. Primary lymphoedema was previously classified purely on age of onset, but the discovery of several causal genes has changed the diagnostic approach, which is now based on clinical phenotyping and genotyping. We now recognise that "primary lymphoedema" is a presenting complaint and not a single condition. There are recognisable patterns of age of onset, distribution, and associated health problems. Primary lymphoedema can be divided into five different categories: 1) lymphoedema associated with syndromic disorders; 2) localised or generalised lymphoedema with systemic/visceral lymphatic abnormalities; 3) lymphoedema in association with disturbed growth and/or cutaneous/vascular anomalies; 4) congenital lymphoedema; 5) late-onset primary lymphoedema. Accurate diagnosis can lead to the identification of the underlying genetic cause, which will lead to better information about the natural history, associated problems, recurrence risks and, hopefully, targeted treatment. The St George's research team have developed a diagnostic algorithm to aid the clinician in their management of patients with primary lymphoedema, and this will be the focus of Dr Gordon's talk.

References:

1) Connell F, Brice G, Jeffery S, Keeley V, Mortimer P, Mansour S. A new classification system for primary lymphatic dysplasias based on phenotype. *Clin Genet* 2010;77(5):438–52.
2) Connell F, Gordon K, Brice G, *et al.* The classification and diagnostic algorithm for primary lymphatic dysplasia: an update from 2010 to include molecular findings. *Clin Genet* 2013;84(4):303–14.