

My Week *Stephen Nutt practises his Welsh*

Monday An early start to the week, made all the more challenging because I was 24 yesterday and celebrated in time-honoured fashion. It's Rare Disease Day on Sunday and we are holding three parliamentary receptions to mark the occasion. We're campaigning for a strategy to address the unmet healthcare needs of people with rare diseases.

Tuesday An even earlier start than yesterday to catch a plane to Belfast for today's reception at the Northern Ireland Assembly. The Cavan Tommy Hoey Trust, one of our members, has been active in getting members of the Legislative Assembly to attend and Debbie, Cavan's mother, gives a moving speech on her son's condition. He has XLP, which affects only 100 families worldwide. Symptoms include severe glandular fever, lymphoma and low levels of immunoglobulins, the proteins in the



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blood that fight infection. There is no recognised cure.

Wednesday Another early start as we finalise our reception at the Welsh Assembly in Cardiff, my home town, tomorrow. I'm looking forward to the opportunity to use my Welsh.

Thursday Flicking through a broadsheet on the train to Cardiff, I come across the story of a family that

volunteered to be a case study for RDUK. Martyn Haden has been living with back pain, leaking spinal fluid and lesions on his neck for five years. Despite having seen many doctors, he has not been able to find an accurate diagnosis. He has had three lots of chemotherapy and has regular CT scans, but his doctors are baffled. Martyn's wife and children practically speak a new language, given the jargon needed to understand his condition. It illustrates the importance of access to information on rare diseases.

Friday The reception went well. Buddug, my colleague in Cardiff, introduced me to some of the families she has been working with. I'm always amazed by people's courage and determination to help others with the same condition.

Rare Disease UK is an alliance of organisations that help people with rare diseases