The Welsh Implementation Plan for Rare Diseases
Consultation Response

From Primary Immunodeficiency UK; www.piduk.org

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Q1 Do you agree that the actions set out relating to empowering those with rare diseases will mean that Wales will meet its commitments?

Yes, in part.

The Local Health Boards are an integral part of the health system in Wales and yet there does not seem to be engagement with patients at this level. It would be good for the LHBs to hold an open annual stakeholder meeting where patients, patient groups etc could input.

PID UK would welcome actions to ensure a cultural change where patient and clinical professionals are seen as equal partners in care. We would also welcome actions that ensure learning and review is based on real patient experience.

To empower patients in Wales it would helpful to define what patient choice means within the Welsh structure for health services.

Q2 Do you agree that the actions set out relating to identifying and preventing rare diseases will mean that Wales will meet its commitments?

Yes in part but it is disappointing that the UK has fallen behind its European counterparts in the number of conditions covered by newborn screening. This is certainly true for primary immunodeficiency (PID) where the technology and know how is already available to screen for rare PIDs, for which effective treatments are available. PID UK would like to see widening of conditions covered by NBS. We would welcome the Welsh Government championing research into the cost/benefit of NBS programmes and making steps to ensure that the patient voice plays an important role in the decisions of the UKNSC for its citizens.

Q3 Do you agree that the actions set out relating to diagnosis and early intervention will mean that Wales will meet its commitments?

Yes, in part.

Diagnosis and early intervention are vital for best patient outcomes and this involves all care level structures, however it is fundamentally how these well these structures interface and work together that will determine patient experience. PID UK would welcome additional actions and evaluation processes to ensure all stakeholders fully interlock and work together effectively. Learning from patient experience is integral to this.

As indicated in the plan some patients may for a long period lack a clear diagnosis or never receive one. For patients and families this is frustrating and stressful. Patients' samples go

into the research pile rather than the diagnostic pathway and patients are left confused as to why the process takes so long and can feel stranded when they are not kept informed. There needs to be greater emphasis at clinic on providing clear information about time lines involved, the process for conditions that are not easily diagnosed, what the tests are and implications for the support of the patient.

PID UK would welcome cross working of rare disease specialists with GPs to design 'flagging' systems that GPs could use in their surgeries to pick up groups of rare conditions. Simple diagnostic criteria could be used to inform the 'flags'.

Next generation sequencing (NGS) will play a huge part in the diagnosis and identification of rare conditions. There needs to be development of educational material for patients/public on NGS and what it means for them. This should be generic but also, as required, covering different groups of conditions. This could be done in liaison with patient support groups.

Q4 Do you agree that the actions set out relating to the co-ordination of care will mean that Wales will meet its commitments?

Yes, in part.

There does not seem to be a mention of firm commitment to the role of a named care co-ordinator within the plan and how this would work. There needs to be greater emphasis on this in the action plan. Adequately resourced care co-ordinators have the potential to make a huge positive effect in ensuring patient best care and experience. It would be helpful for a 'job description' to be developed for this role with the help of patient groups and agencies. This would ensure all stakeholders know how the role works, who will deliver it and what is expected.

Barriers to patient referral such as excessive bureaucracy remain a problem. Streamlining mechanisms that keep the patient and family in the care loop are needed.

Q5 Do you agree that the actions set out relating to the role of research will mean that Wales will meet its commitments?

Yes, in part.

Registries: Rare disease patient registries are extremely important research tools. PID UK would welcome action to develop firm plans, both strategic and financial to ensure the long-term sustainability and extension of patient registries.

Research into rare conditions is vital to improve patient outcomes and yet many patients have little knowledge of how the research process works and how they can participate, and indeed frame the agenda. There does need to be a stronger push to demystify research and clinical trial processes for patients. The actions need to be widened to include how patients will be able to set the future research agenda by strategic priority setting according to their health needs and what they value as outcomes. Actions to produce and deliver a timetable of events for patients in Wales are welcomed.

PID UK welcomes the initiative to develop risk-proportional permission systems (action 39) but challenges remain in streamlining of the ethical review process and bureaucratic processes to ensure there are no unnecessary delays in the start of clinical trials. PID UK would like to see actions to monitor and benchmark this.

When patients become involved in research they expect that the results of the research to be made known to them - transparency and openness are vital and negative, as well as positive, results should be made known. PID UK would welcome actions by the Welsh Government to ensure this happens.

Q6 Do you have any other comments on the draft Plan?

PID UK welcomes the draft plan and the Welsh Government's commitment to its implementation for the benefit of all patients with rare conditions. We look forward to seeing reports on its progress and receiving feedback from patients in Wales on how it is making a difference to their lives.

ENDS.