Cafodd yr ymateb hwn ei gyflwyno i ymgynghoriad y Pwyllgor Iechyd a Gofal Cymdeithasol ar Flaenoriaethau'r Chweched Senedd

This response was submitted to the Health and Social Care Committee consultation on Sixth Senedd Priorities

HSC PSS 126
Ymateb gan: | Response from: Genetic Alliance UK

Blaenoriaethau cychwynnol a nodwyd gan y Pwyllgor
Initial priorities identified by the Committee

Mae'r Pwyllgor wedi nodi nifer o flaenoriaethau posibl ar gyfer ei waith yn ystod y Chweched Senedd, gan gynnwys: iechyd y cyhoedd a gwaith ataliol; y gweithlu iechyd a gofal cymdeithasol, gan gynnwys diwylliant sefydliadol a lles staff; mynediad at wasanaethau iechyd meddwl; arloesi ar sail tystiolaeth ym maes iechyd a gofal cymdeithasol; cymorth a gwasanaethau i ofalwyr di-dâl; mynediad at wasanaethau adsefydlu i'r rhai sydd wedi cael COVID ac i eraill; a mynediad at wasanaethau ar gyfer cyflyrau cronig tymor hir, gan gynnwys cyflyrau cyhyrysgerbydol.

The Committee has identified several potential priorities for work during the Sixth Senedd, including: public health and prevention; the health and social care workforce, including organisational culture and staff wellbeing; access to mental health services; evidence-based innovation in health and social care; support and services for unpaid carers; access to COVID and non-COVID rehabilitation services; and access to services for long-term chronic conditions, including musculoskeletal conditions.

C1. Pa rai o'r materion uchod ydych chi'n credu y dylai'r Pwyllgor roi blaenoriaeth iddynt, a pham?
Q1. Which of the issues listed above do you think should be a priority, and why?

Public Health & Prevention: Expand the Newborn Screening Programme in Wales
Newborn screening plays an important role in aiding the identification of babies at risk of developing rare or genetic conditions, allowing diagnosis before symptoms develop. Such early detection and intervention is particularly important for rare conditions of early childhood, many of which are progressive and irreversible.

Early detection provides an opportunity for stabilising treatment before the deterioration in health or development of a child occurs, potentially preventing the most serious effects of these conditions, which can include severe disability and death. Early diagnosis of an affected child also offers the added benefit of supporting family planning, providing couples with the opportunity to exercise reproductive choices if they wish to.

The Newborn Screening Programme in Wales is not fulfilling its potential. Wales currently screens for just nine conditions, including cystic fibrosis and sickle cell disease. This is far fewer than the current tandem mass spectrometry methods used can detect and far fewer than other comparable high income countries. Iceland,for example, screens for 47 conditions, the Netherlands for 34 and Norway for 28. Decisions on what conditions are screened for in Wales are based on the recommendations of the UK National Screening Committee (UKNSC), although it is important to note that Wales is not bound by these recommendations and could choose to go further.

A comprehensive screening programme in Wales, with the high identification and diagnosis rates it would have the potential to deliver, could contribute vital information to the Welsh Congenital Anomaly and Rare Disease Register and Information Service (CARIS). This will allow our understanding of rare conditions to grow, facilitating research that can lead to future treatments. Some of the newest innovative treatments could not have been developed in Wales, because a screening programme is necessary to recruit children before symptoms arise.

It is for the Welsh Government to make decisions on how the Newborn Screening Programme is delivered in Wales.

The Health and Social Care Committee should consider how the Newborn Screening Programme can be developed and expanded in Wales to improve the detection of rare conditions. Stakeholder groups would include colleagues from the Newborn Screening Division of Public Health Wales, Genetic Alliance UK and a number of patient organisation groups who have an interest in screening for genetic conditions in their communities as well as affected families.

Genetic Alliance UK has produced a Patient Charter on Newborn Screening which can be accessed here: https://geneticalliance.org.uk/wp-content/uploads/2020/01/Newborn-Screening-Report.pdf

Blaenoriaethau allweddol ar gyfer y Chweched Senedd
Key priorities for the Sixth Senedd

C2. Yn eich barn chi, pa flaenoriaethau allweddol eraill y dylai'r Pwyllgor eu hystyried yn ystod y Chweched Senedd mewn perthynas â:

a) gwasanaethau iechyd;
b) gofal cymdeithasol a gofalwyr;
c) adfer yn dilyn COVID?

Q2. In your view, what other key priorities should the Committee consider during the Sixth Senedd in relation to:

a) health services;
b) social care and carers;
c) COVID recovery?

Gwasanaethau iechyd
Health services

Monitor implementation of UK Rare Disease Framework in Wales
1 in 17 people in Wales will be affected by a rare condition at some point in their lives. This equates to approximately 150,000 people.

A condition is considered rare if it affects fewer than 1 in 2,000 people within the general population. Currently, there are over 6,000 known rare diseases.

Many people living with a rare condition in Wales face fundamental challenges in relation to their care. People have reported that some health professionals involved in their care do not know enough about their rare condition to support them. This can result in a delayed diagnosis, a lack of information and missed opportunities to access appropriate care, treatment and research. Care can also be poorly coordinated, contributing to people feeling lost within the healthcare system, unable to effectively manage their condition and, in some particularly concerning situations, failing to receive correct care and treatment.

The implementation of the new UK Rare Diseases Framework provides an opportunity for change. The Framework contains four priorities: helping patients get a diagnosis faster, increasing awareness of rare diseases among healthcare professionals, better coordination of care, and improving access to specialist care, treatment and drugs.
Before the end of 2022, Welsh Government must develop a Rare Diseases Action Plan, which will set out how the priorities identified in the Framework will be addressed.

To successfully deliver the priorities of the Rare Disease Framework, and improve patient outcomes for those living with rare conditions, the Health & Social Care Committee should monitor development and delivery of a Welsh Action Plan to ensure progress has been made post the 2022 deadline.

The Committee might address this issue by scrutinising whether:

-the UK Rare Diseases Framework has been implemented in Wales through the publication of a robust, timely Welsh Rare Diseases Action Plan.

-the infrastructure to collect information on rare conditions across Wales is being developed and can be used to inform service planning.

-there is a central point of information and support for health professionals so that they can better support people with rare conditions.

-appropriate education and training materials for NHS Wales healthcare professionals are being provided

-every patient with a rare condition has access to a care coordinator.

-people living with rare conditions and their families have access to comprehensive, holistic information and support.

The Health and Social Care Committee of the Welsh Parliament could also consider whether a nationally funded rare conditions centre should be developed.

Any work on this topic should be undertaken post March 2022 when the Welsh Action Plan is due to be published.

There is a wide stakeholder group to be consulted on this topic including people affected by rare diseases, including patients and family members, patient group representatives, Genetic Alliance UK, members of the Rare Disease Implementation Group which includes the Chair and leads from each of the Local Health Boards, representatives from the All Wales Medical Genomics Service (both clinical and laboratory), representatives from Welsh Health Specialised Services Committee, Public Health Wales (CARIS and Newborn Screening leads).

The UK Rare Diseases Framework document is available here: https://www.gov.uk/government/publications/uk-rare-diseases-framework

Adfer yn dilyn COVID
COVID recovery

Learnings from Covid-19 to improve the experience of those affected by rare, genetic conditions in Wales

The pandemic caused sudden and for many in the rare, genetic community, devastating changes to their lives. In July 2020, using UK specific data from the EURODIS Rare Barometer Covid-19 Experience Survey and findings from Genetic Alliance UK's weekly community check-ins, the charity produced a report, 'The Rare Reality of Covid-19'. The report highlighted how people living with rare conditions had been placed under immense pressure by the pandemic. Access to appropriate support, information, care and treatments had become more difficult and levels of isolation had increased. The full report can be accessed here: https://geneticalliance.org.uk/wp-content/uploads/2021/08/Annual-Report-201920.-Genetic-Alliance-UK.pdf

In accessing how health services recover from Covid-19, there are a number of recommendations within the report which could be considered by the Committee:
Continuing to deal with Covid-19

– In monitoring the spread of Covid-19, data should be collected that will enable assessment of the impact, in terms of morbidity and mortality, on people living with rare conditions.
Successful transition from crisis state
- For those with rare conditions that confer high risk from Covid-19 who wish to continue protecting themselves through isolation, the support mechanism established should continue to be provided – no one should be forced to stop their protective isolation.
- A clear and short timetable for the reintroduction of services should be published to give people living with rare conditions clarity as to when they can expect a full service from the NHS.
- The provision of remote consultations should be continued. Care should be taken to integrate telemedicine into routine care practice with the necessary clinical assurance and data protection safeguards.
Learning for the future
– The challenges and failures in delivering letters giving shielding advice should be examined, and actions should be taken following this analysis to ensure Wales is able to rapidly provide tailored public health advice to specific populations with specific conditions.