ABMUHB Rare Diseases Delivery Plan 2019/20

Overview A rare disease is defined as a life-threatening or chronically debilitating disease that affects five people or less per 10,000 population.	
Needs Assessment	As a Health Board it is difficult to ascertain the number of patients within our population are that are affected by a rare disease and what exactly their experiences and patient journey has been. With a population of around 530,000 (Stats Wales 2016) and an estimated prevalence for rare diseases of 1 in 17 we estimate around 30,000 of our population may be affected by a rare disease in some way. The Congenital Abnormality Registry (CARIS) produce an update each year of cases reported to CARIS, and prevalence rates of key congenital anomalies and rare diseases which are published as Official Statistics. These figures suggest that Swansea may have a high prevalence rate, but this is thought to reflect high levels of reporting compared to other areas as CARIS office is based in Swansea.
Top 3 Priorities for 2018/19	The Rare disease plan for NHS Wales identifies three priorities in 2018-19, which are unchanged for 2019-20: • Identify and improve the pathway for patients with unknown or delayed diagnosis; • Ensure better use of patient feedback, best practice and evidence to improve pathways for primary, secondary and specialist services; • Improve reporting of rare disease information including epidemiology, significant event analysis and shared learning In 2019-20 the Health Board plans to continue to develop its understanding and focus on Rare diseases and build on its actions in the following ways Continue to build and develop a Local Interest Network to take forward the following actions. Priority 1: To highlight and promote information sharing for rare diseases within the Health Board including using patient stories that were created in 2018-19 Priority 2: To host a local network meeting to bring together stakeholders, and particularly patient representatives to discuss pathway constraints and share learning from patient stories
Outcomes	1. To establish a local connected network to which to bring shared learning and plan improvements where needed
Progress	Progress has been slow with limited resource to take forward However have made progress in:

	Engaging on an All Wales basis to drive better shared working via rare Diseases Delivery Group - Continuing to identifying clinicians and patients with an interest in this area. -
Performance and Audits	TBC
Workforce/Financial	TBC
Condition Specific Cross Cutting/Collaboration	Links with developing genomic strategy
Condition Specific System Shift	

Template 2 – Action Plan 2018/19

Priority Area	Outcome or Target (end 2018/19)	Action	Milestones – in development				Lead Person
			Q1	Q2	Q3	Q4	
Identify and improve the pathway for patients with unknown or delayed diagnosis		To develop a Local Interest Group to take forward actions. To improve performance in relation to quality of spots in Newborn Blood Spot Screening To better understand our local population needs and local pathways					Director of Therapies and Health Sciences
Ensure better use of patient feedback, best practice and evidence to improve pathways for primary, secondary and specialist services		To identify a small number of patients (2-3) with different rare conditions and via face to face interview and case note review: • Complete timeline of events starting from referral made by GP to secondary care • Understand impact this illness had on patient and family • Identify if there are gaps in service provided for a faster diagnosis					Director of Therapies and Health Sciences

		• Identify how care is coordinated • Establish lessons learnt and produce patient stories as a learning device and share learning across practice teams					
Improve reporting of rare disease information including epidemiology, significant event analysis and shared learning		To undertake review of Datix incidents and complaints to identify any issues and/or how we may improve system to identify where feedback is related to rare conditions					Director of Therapies and Health Sciences