SCHEDULE 2 – THE SERVICES

Service Specifications

Service Specification No:	1662
Service	Inherited White Matter Disorders Diagnostic and Management Service (IWMD) (All Ages)
Commissioner Lead	
Provider Lead	

1. Scope

1.1 Prescribed Specialised Service

This service specification covers the provision of an Inherited White Matter Disorders (IWMD) Diagnostic and Management Service. The service will have a small number of separate but collaborating paediatric and adult IWMD Lead Centres that will:

- develop and advise the use of tailored, disease-specific protocols, guidelines, pathways and specialised diagnostics to enable accurate diagnosis and early access to optimal management or treatment, including initiation of treatment trials;
- support local neurology services by providing a diagnosis and management service, including advice on symptom management;
- register patients on the national Rare Disease Registry and the IWMD Service Register; and
- facilitate research including via formal collaboration with other international white matter disease services and registries;
- Ensure that patients and families/carers and are involved in the design and functioning of the network.

1.2 Description

White matter is required for the correct transmission of nerve impulses between neurons involved in cognitive processes such as planning, organising, problem-solving and focussing attention. Inherited White Matter Disorders (IWMD's), also known as leukodystrophies, are a group of rare genetic disorders caused by single gene mutations which affect the white matter in the brain and sometimes the peripheral nervous system (PNS), resulting in clinical presentation with delay or slowing of motor development or loss of previously acquired motor skills. IWMDs comprise a large number of distinct genetic diseases, with over 90 identified to date. Symptoms include visual, gait, feeding/eating difficulties, encephalopathy, seizures and cognitive and psychiatric features, skin conditions, impacts on the bladder and bowel, breathing, hygiene, self-care, sleeping and pain levels.

There is also a gradual deterioration of central nervous system (CNS) function for the majority of patients. Children have an average life expectancy from the point of diagnosis of up to five years, whereas in older patients with symptoms, the progression of the disease tends to be slower and life expectancy longer. The use of novel therapeutics in IWMDs is a rapidly expanding field, particularly in targeting disease specific mechanisms and whilst there are currently no curative treatments for the majority of these diseases, specific treatments may be of benefit to some, siblings can be tested and pre-symptomatic treatment offered if appropriate.

1.3 How the Service is Differentiated from Services Falling within the Responsibilities of Other Commissioners

The service is accessible to all patients of the English NHS with an IWMD. All eligible patients will have access to care and treatment irrespective of their sexual orientation, gender, race, disability, psycho-social circumstances or geographical location. An important feature of all services is that appropriate pathways are developed for socially disadvantaged patients who are often difficult to engage. Access to treatment will be guided by any applicable NHS England national clinical commissioning policies.

1.4 Making a referral

The service will accept inward referrals of patients with suspected or confirmed IWMDs from: tertiary level neurology services; tertiary level metabolic diseases services; clinical genetics services; secondary level services; other specialist services.

1.5 Acceptance Criteria

The service will accept referrals for patients who meet the following criteria:

- Suspected IWMD (including fetal referrals based on antenatal scan and where a sibling has been previously diagnosed with the condition);
- Confirmed IWMD with a specific clinical question concerning management, treatment, or participation in a treatment trial.

1.6 Exclusion Criteria

Treatment such as haematopoietic stem cell therapy (bone marrow transplant) may be offered for these disorders, which are managed jointly by neurology and metabolic diseases services including the national Lysosomal Storage Disorders Service and are outside the scope of this specification.

1.7 Discharge Criteria

In the majority of cases, once the service's multi-disciplinary team (MDT) has agreed the diagnosis and determined any management advice to support local delivery of the individual care plan, clinical care will be wholly provided by local secondary and tertiary services and patients will be discharged to local care. Only complex or undiagnosed cases will be considered for the need for ongoing review.

1.8 Transition

Given the life limiting nature of these genetic conditions, only patients with a slowly progressing version of an IWMD disease, or those whose symptoms present in adolescence will achieve an age to require transition. This is expected to be a small number each year. In such cases, the service will manage transition by setting out clear communications with the most appropriate adult IWMD Lead Unit. Transition planning between the paediatric and adult units will begin at 12 years of age and transfer will take place at 18 years of age.

2. Care Pathway and Clinical Dependencies

2.1 Care Pathway

The service will add value to the patient's pathway by greatly accelerating early diagnosis through rapid access to a virtual diagnostic expert MDT and which will provide optimal interpretation of neuroimaging, next generation molecular genetics and other relevant investigations.
Early diagnosis will enable:

- Optimal management and prognostic advice
- Early recognition and advice on management of co-morbidities.
- Appropriate genetic counselling and life-planning (carrier testing, reproductive choice)
- Initiation of treatment or early participation in a therapeutic trial (when available)
- As appropriate, timely engagement with local palliative care services to ensure good quality end of life care.

Network of Care:
In the IWMD Lead Centres, there will be three levels of assessment based on needs of individual patients:
Level 1: **patients already diagnosed locally**: the expert MDT team will add the details of these patients to a new clinical registry to build up the NHS's understanding of this rare condition and

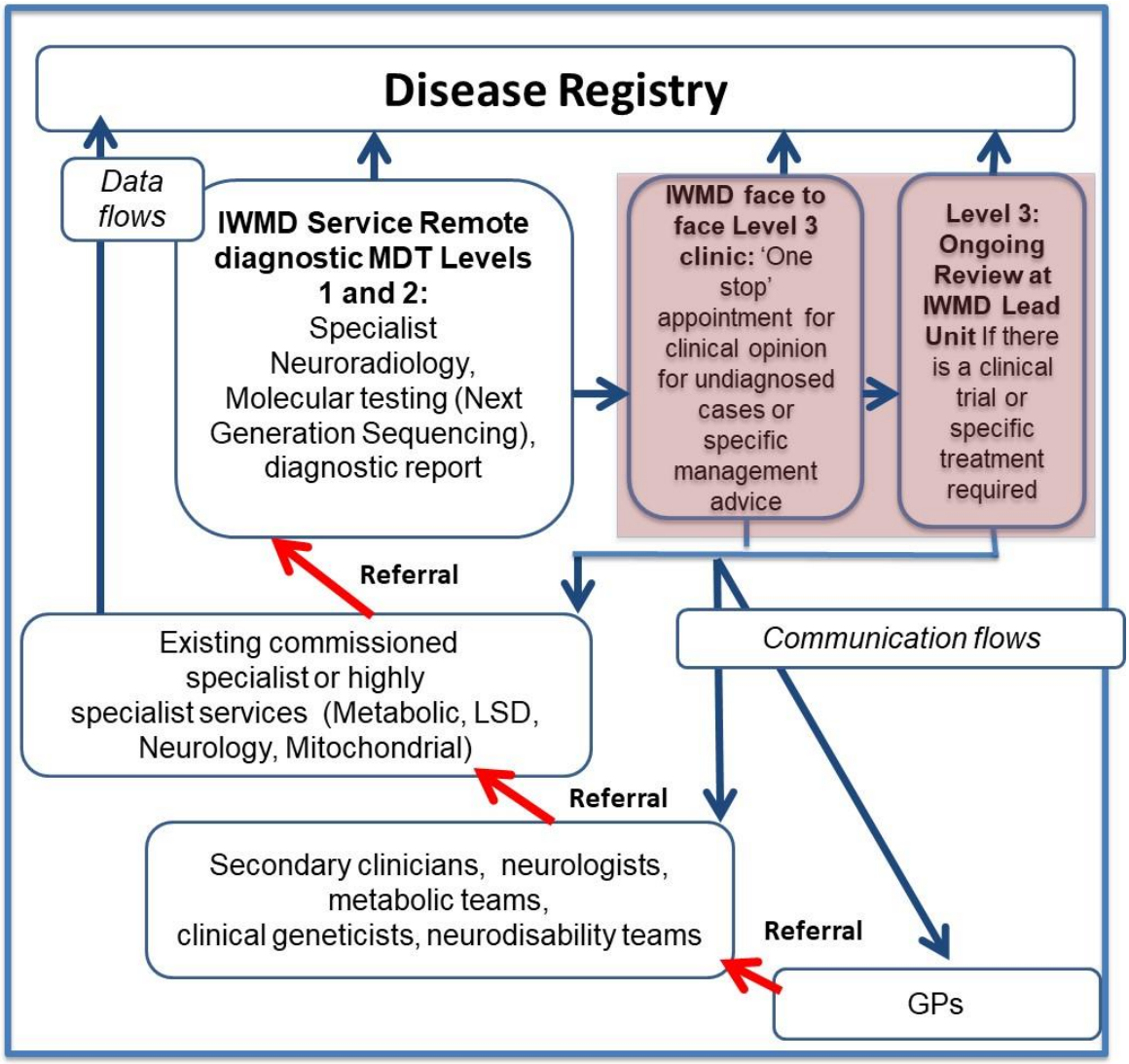
confirm the care plan to reduce the impact of the symptoms on the patient to improve their quality of life.

Level 2: **patients where a diagnosis has not been made**: The remote review MDT will review local test results and make recommendations for further molecular or metabolic testing, review the outcomes of the new tests and advise on best local management.

Level 3: **patients where a diagnosis has not been made and where there is a complexity or concern over management**: the patient will be invited to a clinic in one of the IWMD Lead Centres where there would be a one-stop clinical review by the IWMD service's expert teams and review of test results. Regular review at the IWMD Lead Centres would only occur if the patient was part of a treatment study.

The IWMD Lead Centres will set referral guidelines with referring units to enable ease of communication.

Figure 1: Overview of IWMD Diagnostic and Management Service



Components of service provision

Each of the IWMD Lead Centres will have the following components:

- **Appropriate expertise**: Core service requirements: i) each IWMD Lead Centre will have an appropriate clinical MDT comprising i) consultant neurologist who will be the service's clinical lead, ii) a second named neurologist, iii) a designated consultant neuroradiologist with dedicated

session(s) for reporting MRIs, iv) a designated consultant in clinical genetics with expertise in IWMDs or neurologists with expertise in clinical genetics; v) a designated neurology specialist nurse which will take into account the network role with other IWMD Lead Centres and with referring centres, the patient caseload and clinical commitments; vi) a consultant clinical psychologist or neuropsychologist; vii) a designated consultant in metabolic diseases with expertise in IWMDs; viii) a molecular genetics laboratory scientist with special expertise in IWMDs; ix) an administrative data manager who will also co-ordinate the MDT and support the network and ensure accurate recording of information, and timely communication of decisions to patients and other care providers. IWMD Lead Centres will make provision for succession planning, by having a lead clinician and a further named clinician with an expertise and allocated time in their job plan. The MDT will have a minimum membership of senior skilled staff from the named disciplines on each occasion it meets to be quorate.

- **Appropriate caseload**: each centre will demonstrate that it has a caseload (circa 30 new patients seen in clinic per year per unit) of complex and non-complex patients sufficient to develop and maintain expertise in the diagnosis and management of IWMDs.
- **Access to an MDT for the diagnostic evaluation and/or review of management/treatment issues:** the diagnostic MDT, which may be virtual, will occur jointly with all IWMD Lead Centres; MDT members will have a designated role, although will not need to work exclusively with the service; neurology specialist nurse and clinical psychology support will be available;
- **Access to a designated molecular genetics laboratory:** with expertise in next generation sequencing and in particular IWMDs.
- **Access to neurophysiology**
- **Appropriate clinical support**: access to specialities to support the management of patients with IWMDs; referral to age specific palliative care services; access to appropriate laboratory support (biochemical and pathology) for the diagnosis and management of IWMDs.
- **Appropriate infrastructure:** Each IWMD Lead Centre will have dedicated ring-fenced age-appropriate outpatient clinic facilities: designated conference room facilities with teleconferencing and electronic image viewing facilities (PACs and IEP) and video-conferencing facilities to enable diagnostic MDTs.
- **Robust patient support processes**: provision of written and web-based information; access to welfare advice and support and signposting.
- **Active promotion and delivery of research**, audit, teaching, and training in the area of IWMD's for those working in the IWMD Lead Centres and those working in referring centres. Material, both literature and web based, will be developed for patients and their families/carers, schools or colleges of further education and social services, with age appropriate material where relevant.

2.2 Interdependence with other Services

- In addition to the elements described above, the service will demonstrate that each IWMD Lead Centre has in place:
- access to metabolic services;
- access to an approved molecular genetics service with expertise and infrastructure to perform and interpret whole exome and whole genome sequencing;
- access to neuropsychiatry and age appropriate mental health support (Child and Adolescent Mental Health Services (CAMHS) or adult services);
- access to physiotherapy, occupational therapy, speech therapy, dietetics, local neurodisability services.

3. Population Covered and Population Needs

3.1 Population Covered by this Specification

This specification covers patients of all ages with an IWMD and the population defined as the commissioning responsibility of NHS England. Commissioning arrangements for the devolved nations in relation to this service are as set out in "UK-wide Commissioning Arrangements of Highly Specialised Services" [web link is https://www.england.nhs.uk/publication/nhs-providers-of-highly-specialised-services/

3.2 Population Needs

IWMDs can present at any age however the majority will present in childhood. The mean age of presentation in childhood will be between birth and five years of age and in such cases, there is up to five years life expectancy for patients diagnosed with an IWMD. In adults, the life expectancy for patients with a diagnosis can be up to six years. There is a higher prevalence in some populations in some parts of England.

In the UK, there are likely to be 315 new patients per year (175 children and 140 adults) who present to local services with symptoms. See Appendix A for the list of conditions. Following assessment, any patients who do not have an IWMD will be discharged back to local care, together with a recommendation for onward referral to the most appropriate service if appropriate.

3.3 Expected Significant Future Demographic Changes

Growth levels are expected to be in line with that of the general population.

3.4 Evidence Base

Robust epidemiological data for this group of patients in England does not exist. A single centre United States study from Utah estimated the incidence of IWMDs at 1/7663 births, (Bonkowsy et al Neurology 2010 75:718-25), equating to 106 new patients per year in the UK. Hospital Episode Statistics data for England between the years 2000 - 2010 suggests a prevalence of IWMDs in the 0-19 age range of 5/100,000 population or of 600 cases in total across England which represents the total cohort size. This data is derived from the very broad ICD 10 coding system, and likely to result in missing some cases and including others with a non-IWMD diagnosis. (Fraser L, Department of Health Sciences, University of York. Personal Communication 2013). Based on a recent study in the West Midlands, a total incidence in England of 200 cases per year is suggested, (Wassmer et al 2012 Personal Communication). Although there is an absence of accurate data, given the high rates of genetic disease in certain UK populations/areas, in particular West Yorkshire, the West Midlands and in East London, rates will be higher in these areas and lower elsewhere. Therefore, a figure of 150 cases per year is more realistic.

As for children, there is a lack of epidemiological data for adult onset leukodystrophies. This is compounded by the heterogeneity of these disorders in adults and the overlap with acquired (often treatable) causes which must be thoroughly investigated before a genetic diagnosis is reached. Two recognised adult centres have each been receiving approximately 40 new referrals per year. Given that these centres have not been acting as national referral centres, the incidence is likely higher and estimated total new cases in the UK are approximately 130 to 150 per year (Ahmed et al, 2014 & Lynch et al, 2018 J Neurol Neurosurg Psychiatry. 2014 Jul;85(7):770-81). The recent literature illustrates how next-generation gene sequencing (whole exome and whole genome) has revolutionised the understanding of IWMDs, allowing disease definition and gene identification for numerous rare disorders by focusing on very small groups and individual patients. Knowledge of many new ‘white matter proteins’ is transforming the understanding of white matter physiology and pathophysiology. Early diagnosis and appropriate management are likely to reduce the need for emergency hospital admissions, improve Quality of Life (QoL) and the patient and family experience. Other measures include: Hospital admission data.

Stem cell and gene therapies are evolving rapidly, aiming at personalised therapy for a specific patient with a specific disease. Multimodal approaches targeting multiple aspects of the disease hold the highest promise, (Van der Knaap, MS et al 2016 Neurol Clin Pract 2016;6:506–514). Early diagnosis enables referral for treatments and participation in research studies where appropriate Although incurable, treatment and management is possible for many of the complications and co-morbidities associated with these diseases, (Adang et al 2017 Molecular Genetics and Metabolism Reports 122: 18 -32.

4. Outcomes and Applicable Quality Standards

4.1 Quality Statement – Aim of Service

The aim of this highly specialised service is to provide a diagnostic and management service for patients with an IWMD; minimise inappropriate referrals and avoid any unnecessary travel for the purposes of diagnostic testing and investigations.

It will provide information and support for patients, families and referring clinicians/those professional with whom the patient comes into contact locally; and enable access to novel treatments as they become available. The service will create clinical guidelines for diagnostics and referrals, provide guidance in the event of very rapid deterioration or where an urgent response or amendment to treatment is required, in addition to the provision of patient specific management information sheets. The IWMD Lead Centres will also collaborate with existing local and national experts in regional neuroscience and metabolic disease services and other relevant disease-based national services such as the lysosomal or mitochondrial disease services.

Objectives

To provide a timely service by commissioning a network of existing neuroscience units which have been identified through a provider selection process as having additional expertise in IWMD, which will operate a networked model of care.

NHS Outcomes Framework Domains

Domain 1	**Preventing people from dying prematurely**	✓
Domain 2	**Enhancing quality of life for people with long-term conditions**	✓
Domain 3	**Helping people to recover from episodes of ill-health or following injury**	✓
Domain 4	**Ensuring people have a positive experience of care**	✓
Domain 5	**Treating and caring for people in safe environment and protecting them from avoidable harm**	✓

Outcomes

4.2 Indicators Include:

Number	Indicator	Data Source	Outcome Framework Domain	CQC Key question
Clinical Outcomes				
101	Proportion of patients receiving an MDT discussion	Provider / SSQD	1, 5	Safe, effective, caring, responsive
102	Proportion of level 2 patients who receive next generation sequencing molecular testing	Provider / SSQD	1,5	Safe, effective, caring, responsive
103	Proportion of level 3 clinics where key MDT staff groups as outlined in the service specification are represented and meet with the patient.	Provider / SSQD	1, 5	Effective, caring
104	Proportion of parents of a paediatric IWMD patient who receive genetic counselling.	Provider / SSQD	4, 5	Effective, caring

105	Proportion of adult IWMD patients who receive genetic counselling.	Provider / SSQD	4, 5	Effective, caring
106	Proportion of cases in which a treatment plan has been agreed.	Provider / SSQD	1, 2, 3	Effective, caring
107	Proportion of patients recruited to a clinical trial	Provider / SSQD	1,2	Effective, caring
Patient Experience				
201	The IWMD Lead Centre has a strategy/plan in place for engaging with patients, families/carers and stakeholder groups.	Self declaration	2, 3, 4, 5	Effective, caring, responsive
202	There is patient information leaflet for inherited white matter disorders as per the service specification.	Self declaration	5	Effective, caring
203	The IWMD service undertakes a patient and family/carer experience exercise at least annually.	Self declaration	2, 5	Effective, caring, responsive
Structure and Process				
301	The IWMD Lead Centre has a multi-disciplinary team as detailed within the service specification.	Self declaration	1, 2, 3, 4, 5	Safe, effective
302	The IWMD Lead Centres lead on actively promoting and delivering research, audit, teaching, and training in the area of IWMDs.	Self declaration	1, 2, 3, 4, 5	Safe, effective, well led
303	There are virtual multi-professional MDT meetings to consider level 1 and level 2 patients at least monthly unless the meeting falls on a public holiday	Self declaration	1, 2, 3, 4, 5	Safe, effective, caring, well led
304	The IWMD Lead Centres will hold multi-disciplinary team-led Level 3 clinics at least quarterly.	Self declaration	1, 2, 3, 4, 5	Safe, effective, caring, well led
305	There are clinical guidelines in place as detailed within the service specification	Self declaration	1, 2 ,3	Safe, effective
306	There are patient pathways in place as detailed within the service specification	Self declaration	1, 2, 3, 4	Safe, effective
307	The team participates in national and local audits.	Self declaration	2, 3, 4	Safe, effective, well led
308	The team submits data to the National Database (National Disease Registry)	Self declaration	2, 4	Safe, effective, well led

See also Appendix B for full description of each indicator. See also Schedule 6 of the contract for detailed definitions of indicators.

4.3 Commissioned providers are required to participate in annual quality assurance and collect and submit data to support the assessment of compliance with the service specification as set out in Schedule 4A-C. All IWMD Lead Centres must hold and attend joint national annual clinical meetings attended by the Highly Specialised Service commissioning team. IWMD Lead Centres and their referring units will contribute to national data collection of treatment and outcomes and support development of a national database. Each IWMD Lead Centre must ensure that: all practitioners participate in continuous professional development and networking; patient outcome data is recorded and audited across the service; it participates in the national audit commissioned by NHS England, including the collection of experience and outcome data

from their referrers. Annual clinical meetings will address: clinical performance and outcomes; process-related indicators e.g. efficiency of the assessment process, prescribing policy, bed provision and occupancy, outpatient follow-up etc; stakeholder satisfaction, including feedback from patients, families, referring clinician and GPs.

4.4 Applicable CQUIN goals are set out in Schedule 4D

To be agreed with the commissioner.

5. Applicable Service Standards

5.1 Applicable Obligatory National Standards

There are no NICE standards specifically for this patient group. The service will be commissioned to support the aims, objectives and commitments of the UK Strategy for Rare Disease.

5.2 Other Applicable National Standards to be met by Commissioned Providers

To be established.

5.3 Other Applicable Local Standards

This service specification will ensure rapid access to specialist clinical, radiological and genetics expertise. This will enable rapid diagnosis, early recognition of co-morbidities, appropriate counselling and life-planning and, for some patients, initiation of treatment or participation in a therapeutic trial. The providers of the service must ensure they are fully integrated into their Trust's corporate and clinical governance arrangements and comply fully with the Clinical Negligence Scheme for Trusts (CNST) and Care Quality Commission (CQC) requirements in terms of quality and governance.

6. Designated Providers (if applicable)

The IWMD Diagnostic and Management Service will comprise a small number of IWMD Lead Centres (up to three paediatric units and two adult units), located in the North, Midlands and London/the South which will develop good relationships with referring centres. IWMD Lead Centres will be defined through the compliance process on the basis of pre-existing specialist neurology expertise. Depending on the outcome of the procurement process, the IWMD Lead Centres may be multi-sited. These will meet in a national co-ordinating group up to four times per year to discuss audit, learning and the findings from diagnosing and managing the patient cohort.

7. Abbreviation and Acronyms Explained

The following glossary contains the abbreviations and acronyms which have been used in this document:

Glossary of Terms

CNS	Central Nervous System	The nervous system is made up of two main components: the central nervous system (CNS) and the peripheral nervous system (PNS): The CNS comprises the brain and spinal cord.
DNA	Deoxyribonucleic Acid	A self-replicating material which is present in nearly all living organisms as the main constituent of chromosomes. It is the carrier of genetic information. Changes in the DNA (mutations) are the cause of genetic diseases.
ICD 10	International Statistical Classification of Diseases and Related Health Problems (ICD), 10th revision	A medical classification list by the World Health Organisation (WHO). It contains codes for diseases, signs and symptoms, abnormal findings, complaints, social circumstances, and external causes of injury or diseases.

LSD	Lysosomal Storage Disorders	Genetic diseases of lysosomes which are organelles in the cells in the body. Some of the LSDs result in an IWMD however many IWMDs are not LSDs. LSDs are already a nationally commissioned service
MDT	Multi-Disciplinary Team	A Multidisciplinary Team/meeting is a meeting of the group of professionals from one or more clinical disciplines who together make decisions regarding recommended treatment of individual patients.
MR, MRI	Magnetic Resonance Imaging	A scanning procedure which uses strong magnetic fields and radio waves to produce detailed images of the inside of the body.
	Neurodisability	Neurodisability is an umbrella term for conditions associated with impairment of the nervous system.
NGS	Next Generation Sequencing	The use of technology that enables large volumes of genetic material (DNA) to be analysed in a single test from a single sample
PNS	Peripheral Nervous System	The peripheral nervous system is the part of the nervous system that consists of the nerves and ganglia outside of the brain and spinal cord.
PACs	Picture Archiving and Communications System	PACS enables x-ray and scan images to be stored electronically and viewed on screens, helping to improve diagnosis methods
White Matter	WM	The white matter is one of the major components of the brain and spinal cord. It has many important roles in normal brain development and function and diseases of the white matter result in impaired brain function resulting in gait abnormalities, learning difficulties, feeding difficulties, weakness, visual problems and early death.

References

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Appendices:

Appendix A: List of leukodystrophies currently recognised (not exhaustive);

Appendix B: Full description of quality metrics

Appendix A: List of IWMD's/Leukodystrophies currently recognised

Title of clinical condition	Other names	Gene if known
AARS2 related leukodystrophy	Ovario leukodystrophy	*AARS2*
Adult onset autosomal dominant leukodystrophy	Lamin related LD	*LMNB1*
AGC1 deficiency associated hypomyelination	AGC1	*SLC25A12*
Aicardi-Goutieres syndrome		*TREX1* *RNASEH2A* *RNASEH2B* *RNASEH2C* *SAMHD1* *ADAR1* *IFIH1*
AIMP1 mutation related hypomyelination		*AIMP1*
Alexander disease		*GFAP*
Canavan disease		*ASPA*
Cathepsin A related arteriopathy with strokes and leukoencephalopathy	CARASAL	*CTSA*
Cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopahty	(CADASIL)	*NOTCH3*
Cerebral autosomal recessive arteriopathy with subcortical infarcts and leukoencephalopathy	(CARASIL)	*HTRA1*
Cerebrotendinous xanthomatosis		*CYP27A1*
CLC2 related disease		*CLC 2*
Coats Plus disease		*CTC1* *POT1* *STN1*
Fucosidosis		*FUCA1*
Giant axonal neuropathy		*GAN*
Hereditary diffuse leukoencephalopathy with spheroids	(HDLS)	*CSF1R*
Hypomyelination with atrophy of the basal ganglia	HABC	*TUBB4A*
Hypomyelination with congenital cataract	HCC	*HYCCIN (FAM126A)*
Hypomyelination with brainstem and spinal cord involvement and leg spasticity	HBSL	*DARS*
HSPD related hypomyelination		*HSPD1*
Krabbe leukodystrophy	Globoid cell leukodystrophy	*GALC*
Leighs disease		(many genes)
Leukoencephalopathy with brainstem and spinal cord involvement and high lactate	LBSL	*DARS2*
Leukoencephalopathy with calcification and cysts	Labrune disease	*SNORD118*
Leukoencephalopathy with thalamus and brainstem involvement and elevated lactate	LTBL	*EARS2*
Megalencephalic leukosdystrophy with subcortical cysts	MLC	*MLC1* *GLIALCAM*
Metachromatic leukodystrophy		*ARSA*
Mitochondrial disease associated leukodystrophies		Many different genes
Nasu Hakola disease	Polycystic lipomembranous osteodysplasia with	*TREM2* *TYROBP*

	sclerosing leukoencephalopathy (PLOSL)	
Oculodentodigital dysplasia		*GJA1*
Pelizaeus-Merzbacher disease		*PLP1*
Pelizaeus - Merzbacher like disease		*GJC2* *SOX10* *MAG*
Peroxisomal disorders (many genes)		*PEX* genes
Polyglucosan body disease		*GBE1*
POLR3 related leukodystrophy	4H syndrome Tremor ataxia with central hypomyelination (TACH)	*POLR3A* *POLR3B*
POLR1C related leukodystrophy		*POLR1C*
RARS related hypomyelination	RARS	TBA
Sialic acid storage disorders		*SLC17A5*
Sjogren-Larsson syndrome		*ALDH3A2*
Vanishing white matter disease	Childhood ataxia with CNS hypomyerlination (CACH)	*EIF2B1 -5*
X-linked adrenoleukodystrophy and adrenomyeloneuropathy		*ABCD1*

***** For some of the above there are multiple specific genetic subtypes e.g. mitochondrial or peroxisomal disorders

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Appendix B: Full description of quality metrics

Indicator		Detail			Data Source	O.F Domain: 1,2,3,4,5	CQC Key question: Well led, responsive, effective, caring, safe
Number		Descriptor	Notes	Evidence documents			
Clinical Outcomes - quantitative data where possible using national data need to minimise the burden							
101	Proportion of patients who receive a full MDT discussion.	Proportion of patients who receive a full MDT discussion.		Annual Report	Provider / SSQD	1, 5	Safe, effective, caring, responsive
102	Proportion of level 2 patients who receive next generation sequencing molecular testing.	Proportion of level 2 patients who receive next generation sequencing molecular testing.		Annual Report	Provider / SSQD	1, 5	Effective, caring
103	Proportion of level 3 clinics where key MDT staff groups as outlined in the service specification are represented and meet with the patient.	Proportion of level 3 clinics where staff groups as outlined in the service specification are represented and meet with the patient.		Annual Report	Provider / SSQD	1, 5	Effective, caring
104	Proportion of parents of a paediatric IWMD patient who receive genetic counselling.	Proportion of parents of a paediatric IWMD patient who receive genetic counselling.		Annual Report	Provider / SSQD	4, 5	Effective, caring
105	Proportion of adult IWMD patients who receive genetic counselling.	Proportion of adult IWMD patients who receive genetic counselling.		Annual Report	Provider / SSQD	4, 5	Effective, caring
106	Proportion of cases in which a treatment plan has been agreed.	Proportion of cases in which a treatment plan has been agreed.		Annual Report	Provider / SSQD	1, 2, 3	Effective, caring
107	Proportion of patients recruited to a clinical trial.	Proportion of patients recruited to a clinical trial.		Annual Report	Provider / SSQD	1,2	Effective, caring
Patient Experience - PROMS PREMS can be difficult to gather if no national survey can put in process indicator if required							

201	The IWMD Lead Centre has a strategy/plan in place for engaging with patients, families/carers and stakeholder groups.	The IWMD Lead Centre has a strategy/plan in place for engaging with patients, families/carers and stakeholder groups. There is a plan detailing how to increase patient/parent involvement in the design and functioning of the network.		Operational Policy	Self declaration	2, 3, 4, 5	Effective, caring, responsive
202	There is patient information leaflet for inherited white matter disorders as per the service specification.	There is patient information that includes: - details relating to the condition; - patient pathway details including treatment details; - national and local support groups; - emergency contact details.		Operational Policy	Self declaration	5	Effective, caring
203	The IWMD service undertakes a patient and family/carer experience exercise at least annually.	The IWMD service undertakes a patient and family/carer experience exercise at least annually, reviewing the feedback and implementing any actions as appropriate, and feeds back to patients/carers.		Annual Report	Self declaration	2, 5	Effective, caring, responsive
Structure and Process - infrastructure requirements, staffing, facilities etc.							

301	The IWMD service has a multi-disciplinary team as detailed within the service specification.	The IWMD service has a multi-disciplinary team as detailed within the service specification and includes: a consultant neurologist who will be the clinical lead for the service; a second named neurologist; a designated consultant neuroradiologist with dedicated session(s) for reporting MRIs; a designated consultant in metabolic diseases with expertise in IWMDs; a designated consultant in clinical genetics with expertise in IWMDs; a designated neurology specialist nurse; a consultant clinical psychologist or neuropsychologist; a data manager who will also co-ordinate the MDT and support the network. The nursing role will take into account the network role with other IWMD Lead Centres and provide a link with referring centres.		Operational Policy	Self declaration	1, 2, 3, 4, 5	Safe, effective
302	The IWMD Lead Centres lead on actively promoting and delivering research, audit, teaching, and training in the area of IWMDs. There is a process whereby the IWMD Lead Centre reviews issues relating to governance and outcomes.	The IWMD Lead Centre has a process in place for enabling the review of outcomes, audit activity, research and training. The process details the steps taken in relation to review and how communication with referring centres is achieved.		Operational Policy	Self declaration	1, 2, 3, 4, 5	Safe, effective, well led

303	There are multi-professional MDT meetings to consider level 1 and level 2 patients held at least monthly which include all staff disciplines listed in the service specification.	There are multi-professional virtual MDT meetings for level 1 and level 2 cases held at least monthly, and these include input from: Consultant Neurologist; Consultant Neuroradiologist; Neurology Specialist Nurse; Metabolic Consultant; Clinical Geneticist; Administrator, Data Manager. Attendance of MDT members and treatment decisions are recorded accordingly. There should be quorate meetings of the MDTs on at least 95% of the scheduled dates.		Operational Policy / Attendance records including quoracy	Self declaration	1, 2, 3, 4, 5	Safe, effective, caring, well led
304	There are multi-disciplinary team-led level 3 clinics in place including the disciplines listed in the service specification.	There are clinics including MDT input from the following professionals: Consultant Neurologists; Consultant Neuroradiologist; Metabolic Consultant; Neurology Specialist Nurse; Psychologist; and any other disciplines as required.		Operational Policy	Self declaration	1, 2, 3, 4, 5	Safe, effective, caring, well led
305	There are clinical guidelines as detailed within the service specification.	There are clinical guidelines in place, which, where available, reflect national guidelines.		Operational Policy	Self declaration	1, 2 ,3	Safe, effective
306	There are patient pathways as detailed within the service specification.	There are patient pathways as detailed within the service specification.		Operational Policy	Self declaration	1, 2, 3, 4	Safe, effective
307	The team participates in national and local audits.	The team participates in national and local audits, meeting with the NHS England Highly Specialised Service team and sharing/reviewing clinical activity and outcomes at least annually.		Operational Policy / Annual Report	Self declaration	2, 3, 4	Safe, effective, well led

308	The team submits data to the National Database (National Disease Registry).	The team submits data to the National Database (National Disease Registry).		Operational Policy	Self declaration	2, 4	Safe, effective, well led