**Genetic disorders in the Irish Traveller population**

Irish Travellers are an endogamous, ethnically Irish population numbering approximately 40,000 within the island of Ireland with up to 10,000 living on mainland Europe. Irish Travellers practice cousin marriage and we recognise > 90 autosomal recessive disorders that occur within their population. The majority of autosomal recessive disorders are due to homozygous mutations. Certain disorders cluster within certain clans and there are geographical differences in incidence of disorders depending where they come from in Ireland. There is expertise within our group on the disorders common within this population which could help with diagnosis in children and adults presenting to centres where the expertise does not exist.


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**Clinical Laboratory Network Members**
Molecular Laboratory, Department of Genetics

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Academic Centre for Rare Disease, University College Dublin  
Dr Jillian Casey  
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We provide expert advice to clinicians as needed – please contact Dr. Lynch, network co-ordinator.
Top diagnoses in Metabolics:  
Galactosemia  
Hurler Syndrome  
I cell disease  
Hyperprolinaemia  
Infantile liver failure type 1 ILFS1  
Glutaric aciduria type 1  
Glycogen storage disorder (GSD) types III and V  
Classical PKU and pterin defects  
Methylmalonic aciduria  
Leigh disease and Respiratory Chain Disorders  
Sly syndrome

Top diagnoses in Clinical Genetics  
Osteogenesis Imperfecta (lepre1 gene)  
Natural Killer Cell and Glucocorticoid deficiency with DNA repair defect NKGCD  
Primary ciliary dyskinesia (Genetic Heterogeneity)  
McArdles  
Microcephaly  
Fanconia’s anaemia (Genetic Heterogeneity)  
Walker-Warburg  
Merosin negative myopathy  
Multiple epiphyseal dysplasia  
Cardiomyopathy  
Deafness  
Cohen syndrome  
Lebers amaurosis (Genetic Heterogeneity)

Top diagnoses in neurology  
Epilepsy  
Friedreich’s Ataxia  
I cell disease  
Leigh disease  
GSD V  
Microcephaly  
Walker-Warburg  
Merosin negative myopathy  
Lebers amaurosis  
Cohen syndrome

Top diagnosis in endocrinology  
Natural Killer Cell and Glucocorticoid deficiency with DNA repair defect NKGCD  
Congenital adrenal hyperplasia  
46,XY female phenotype (Genetic Heterogeneity)  
Osteogenesis imperfecta  
Familial Hyperinsulinism (Genetic Heterogeneity)  
Wolcott-Rallison