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.

For more information about genetic disorders among Irish Travellers please consult our recent publication: ***Catalogue of inherited disorders found among the Irish Traveller population;*** Lynch SA, Crushell E, Lambert DM, et al. J Med Genet Epub ahead of print: [12 Feb 2018]. doi:10.1136/jmedgenet-2017-104974

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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