Despite early diagnosis through Newborn Bloodspot Screening and strict dietary treatment, there are long-term complications of Classical Galactosaemia, including female infertility, osteopenia, and, in some cases, learning disabilities or neurological symptoms such as tremor or ataxia. Therefore, neurological symptoms may easily be attributed to an underlying diagnosis of Classical Galactosaemia. However, the coexistence of classical Galactosaemia and Friedreich’s Ataxia (FRDA) was previously reported amongst the Irish Traveller population. Classical Galactosaemia and FRDA are both autosomal recessive conditions, the gene loci for which are located on either side of the centromere of chromosome 9. FRDA is one of the most common forms of autosomal-recessive ataxia and slowly progressive; it also commonly involves the heart. We here present the clinical spectrum of 10 Irish patients currently being treated in our Metabolic Centre in whom a diagnosis of Classical Galactosaemia together with FRDA was made. All patients were diagnosed with Classical Galactosaemia through Newborn Screening first. All were diagnosed with the common Classical Galactosaemia GALT mutation Q188R/Q188R. Eight of the ten patients later presented with progressive ataxia, between the ages of 7–14 years. One child presented in cardiac failure secondary to dilated cardiomyopathy at 7 years of age. It was noted that he was not ataxic at presentation of FRDA and that he had normal tendon reflexes. Another patient who was diagnosed at 6 years of age still had an essentially normal neurological exam almost 4 years later. The diagnosis of FRDA was confirmed by detecting the common pathogenic GAA repeat expansion mutations in both alleles of the frataxin gene (FXN) in affected individuals.

Taken together, neurological signs may easily be attributed to an established diagnosis of Classical Galactosaemia. However, one should be vigilant for the coexistence of Classical Galactosaemia and FRDA in a high-risk population, particularly when monitoring patients with a complex phenotype, such as a neurological or cardiac presentation.

Objective To examine the neuropsychological profile of children at perinatal risk with or without CP, compare them to each other and to the normative group. It is expected that more specific cognitive deficits will be found in both clinical groups in relation to the normative group, as well as more deficits in children with CP compared to children at perinatal risk without CP.

Results Both clinical groups had significantly lower results in 12 out of 13 measures applied in relation to the normative group: in Auditory and Visual Attention, Speeded Naming, Comprehension of Instructions, Fingertip Tapping, Imitation Hand Positions, Visuomotor Precision, Design Copy, Arrows, Memory for Faces, Memory for Names and Narrative Memory (z-score = -.29 to -1.67 in the CP group and z-score = -.38 to -1.07 in the group at risk). The CP group had lower results in comparison to the group at risk in 5 measures which include a motor component (oral or manual) in performance. The group at risk had worse results in the Memory for Names than the CP group.

Conclusion The children at perinatal risk achieved significantly lower neuropsychological results in comparison to the normative group, regardless of motor deficit (in the most of measures) or any demographic and perinatal variable or EEG findings. It may be concluded that both clinical groups are at risk for specific cognitive deficits. That has significant implications for neuropsychological examination and treatment, where the model of developmental neuropsychological psychotherapy may be applied.