

PMM2-CDG is the most common of a growing family of more than 100 extremely rare inherited metabolic disorders. More than 800 cases of this specific disorder have been reported worldwide. Two other disorders in this family are each represented by more than 20 cases. The remaining disorders in this family have fewer than 20 cases with several earning a place in the medical literature based on the report of one or two cases. The exact incidence and prevalence of these disorders in the general population is unknown. It is difficult to determine the true frequency of PMM2-CDG in the general population since the disorder may still be under-recognized and under-diagnosed in many parts of the world.