### Background

The co-existence of two distinct disorders, Phenylketonuria (PKU) and Myasthenia Gravis (MG) in the same patient has rarely been reported.

PKU is an inborn error of the metabolism resulting from a phenylalanine hydroxylase deficiency. MG is a chronic autoimmune disorder in which antibodies destroy the communication between nerves and muscle, resulting in weakness of the skeletal muscles.

# **Case presentation**

We report a case of a 16-year-old girl affected by classic PKU and MG. She had growth failure, poor skin pigmentation, microcephaly, seizures, and global developmental delay. The PKU was detected by elevated levels of phenylalanine, and confirmed by the molecular data at the age of 1.5 years. The MG was suspected at the age of 14 years on the presence of gait problems, easy fatigability, ptosis, waddling gait, reduce muscle force in upper and lower extremities(3/5), and difficulty in swallowing and climbing the stairs which were apparently not related to PKU. The MG was confirmed by the positive anti-acetylcholine receptor (AChR) antibody and positive RNS (repetitive nerve stimulation) tests.

After corticosteroid and Pyridostigmine initiation her symptoms recovered. Now she is on Pyridostigmine twice a day and she is symptom free.

# The Co-existence of Phenylketonuria and Myasthenia Gravis: A Case Report

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	Name	Phenotype	Exons	Genotype	Informative	VNTRs	Sequencing	MPLA
					RFLPs			
	Father	Carrier	3	deletion in exon 3/n	PVUII,	Informative	~	$\checkmark$
					BGLII			
/	Mother	Carrier	6	CD198 Small deletion /N	PVUII, BGLII	Informative	✓	
9	Patient	Affected	3, 6	CD198 Small deletion /deletion in exon 3				•



р.	Decr. (%)	Area	Decr. (%)
l mV	0.0	92.3 mVms	0.0
LmV	19.6	65.3 mVms	29.2
2 mV	33.4	53.4 mVms	42.2
l mV	42.8	45.2 mVms	51.0
LmV	44.2	43.8 mVms	52.6
δmV	44.8	44.1 mVms	52.2
8 mV	41.9	47.0 mVms	49.0
) mV	40.2	48.0 mVms	48.0
l mV	41.8	46.6 mVms	49.5

### Conclusion

The co-existence of PKU and MG, due to the prevalence of each disease, could be a rare event. Since some symptoms of PKU and MG overlap, it is very important that the possibility of their coexistence be kept in mind for the proper management of both diseases.

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# References

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