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# Pallister-Killian Mosaic Syndrome

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## INTRODUCTION

Pallister-Killian Mosaic Syndrome (PKMS) is a very rare, multiple congenital developmental disorder of unknown prevalence that is characterized by hypotonia, intellectual disability, seizures, distinctive facial features, meagre hair, unusual skin pigmentation, and other birth defects. Incidence is around 1/25,000. They often have difficulty breathing, feeding, sitting, standing, walking, and speech. The facial features include a high, round forehead, broad nose bridge, telecanthus, a wide mouth and a large tongue. Hearing loss, vision impairment, genital abnormalities, and heart defects are frequently noted. The genetic etiology is a result of isochromosomes that have either two q arms (long) or two p arms (short). Therefore isochromosome 12p is a chromosome 12 with two p arms. Most cells have two copies of each chromosome with one from each parent. Pallister-Killian mosaic syndrome, cells have the two usual copies of chromosome 12, but also have the isochromosome 12p. This results in a total of four copies of all the genes on the p arm of chromosome 12 which causes a disruption in the normal development, resulting in the characteristic features of PKMS. Only about 100 cases have been reported in the literature.

## CASE REPORT

SS is a 4 year 4 month old female with PKMS who presents for an evaluation of eye rubbing in both eyes. Her parents are interested in determining if glasses and/or vision rehabilitative therapy are needed. She is currently receiving OT, PT, speech and vision services. SS was taking no systemic medications and denied any allergy to medication. Her last evaluation noted a moderate amount of hyperopia and astigmatism. She has been wearing this prescription for about 2 months. Her father reports that she wears them without difficulty, but does not notice any improvement in her visual abilities.

Her visual acuities were variable (fix and follow). Pupils were equal, round and sluggishly reactive to light. The Bruckner test showed OD slightly whiter and brighter, Hirschberg: >40 AXT and Kappa: central, but unsteady fixation OD/OS. EOMs appeared full. Her cycloplegic refraction noted OD +4.50 -1.00 X180 and OS +4.50 -1.00 X 090. The external health assessment revealed a petechial hemorrhage OD and was otherwise unremarkable. The DFE was also unremarkable. SS fell asleep early into this examination.

## SPECIAL TESTING

A visually evoked response (see below) was completed at a later date. A bright flash VEP OU noted that the waveform was well formed, P120= 12.56uV, 168ms (variable latency between 154-168 ms); OD a well formed waveform, p120= 11.3uv, 162ms (variable latency between 162-170ms) and OS showed a more variable wave form (patient was getting tired), P120=6.77uV, 126ms (latency was between 110-126). It was difficult to determine at this time which eye was seeing better since the OS latency was better but the amplitude OD was larger. The VEP showed a cortical response to light but the limitations and variability seen is consistent with a diagnosis of cortical visual impairment.

## ASSESSMENT

The assessment included pediatric cortical visual impairment, exotropia, and allergic conjunctivitis as well as hyperopia and astigmatism.

## PLAN

The plan is to wear her glasses full time, prescribe Pataday for the ocular allergy present, repeat the VEP in about 12 months and institute vision rehabilitation/vision stimulation therapy.

## CONCLUSION

Those with pediatric cortical visual impairment have been shown to improve in vision function once an appropriate diagnosis has been made and intervention given. Since Pallister-Killian Mosaic Syndrome is rare we do not know if this is also true for PKMS. This poster discussed all additional assessments and the possible outcomes of a vision rehabilitation/vision stimulation program.

## REFERENCES

Tilton RK, Wilkens A, Krantz ID, Izumi K. Cardiac manifestations of Pallister-Killian syndrome. *Am J Med Genet A.* 2014 May;164A(5):1130-5. doi: 10.1002/ajmg.a.36413. Epub 2014 Feb 6. PMID:24504854

Wilkens A, Liu H, Park K, Campbell LB, Jackson M, Kostanecka A, Pipan M, Izumi K, Pallister P, Krantz ID. Novel clinical manifestations in Pallister-Killian syndrome: comprehensive evaluation of 59 affected individuals and review of previously reported cases. *Am J Med Genet A.* 2012 Dec;158A(12):3002-17. doi: 10.1002/ajmg.a.35722. Epub 2012 Nov 20.

Candee MS, Carey JC, Krantz ID, Filloux FM. Seizure characteristics in Pallister-Killian syndrome. *Am J Med Genet A.* 2012 Dec;158A(12):3026-32. doi: 10.1002/ajmg.a.35567. Epub 2012 Nov 20.

Birch M, Patterson A, Fryer A. Hypopigmentation of the fundi associated with Pallister-Killian syndrome. *J Pediatr Ophthalmol Strabismus.* 1995 Mar-Apr;32(2):128-31.

Graham W, Brown SM, Shah F, Tonk VS, Kukulich MK. Retinal pigment mosaicism in Pallister-Killian syndrome (mosaic tetrasomy 12p). *Arch Ophthalmol.* 1999 Dec;117(12):1648-9.

Genetics Home Reference: <http://ghr.nlm.nih.gov/condition/pallister-killian-mosaic-syndrome>

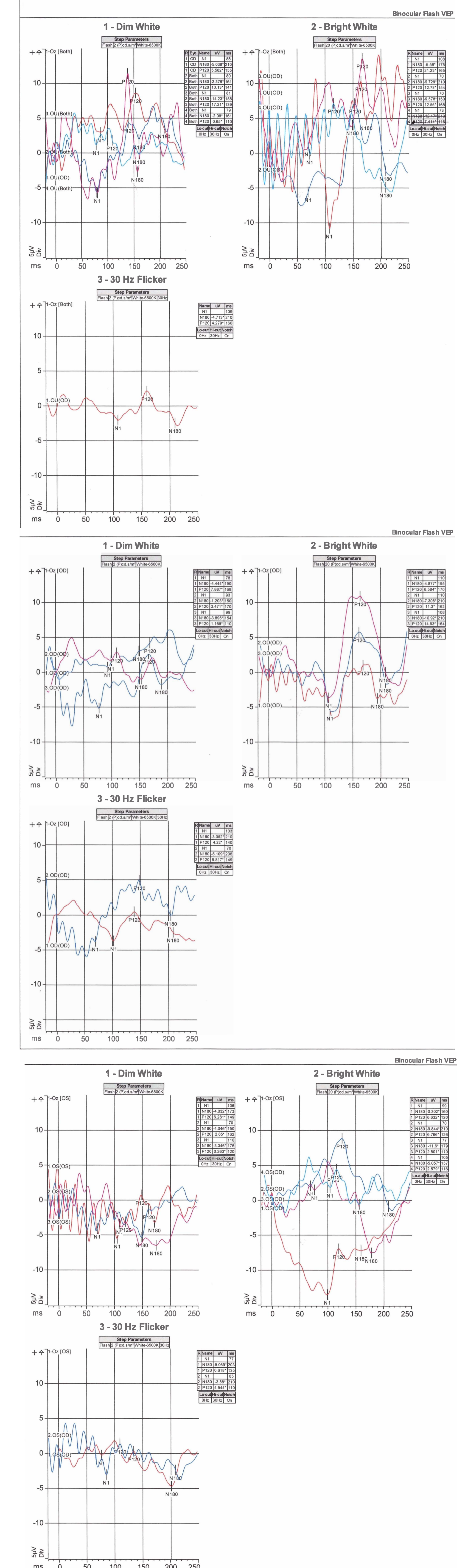
## ORGANIZATIONS

PKS Kids: <http://pkskids.net/>

NORD: <https://www.rarediseases.org/rare-disease-information/rare-diseases/byID/512/viewAbstract>

OMIM: <http://omim.org/entry/601803>

## VEP Outcomes OU, OD and OS



## Characteristics of Pallister-Killian Mosaic Syndrome

Motor	Cognitive	Facial	Sensory	Genetics	Etiology	Other Names	Other
Hypotonia	Severe/ Profound Intellectual Disability	high, rounded forehead	hearing loss	Not inherited	caused by the presence of an abnormal extra chromosome	isochromosome 12p syndrome	extra nipples
Breathing problems		broad nasal bridge	Visual impairment			PKS	genital abnormalities
Speech Delay		short nose				Teschler-Nicola/Killian syndrome	heart defects
Sitting, Standing, Walking Delay		Telecanthus				tetrasomy 12p, mosaic	skeletal abnormalities
Feeding problems		low-set ears					short arms and legs
		rounded cheeks					congenital diaphragmatic hernia
		thin upper lip					iris transillumination defects
		cleft palate					Retinal pigment anomalies

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