

## Case Study 6: 8 month old boy with Joubert's Syndrome and significant visual impairment due to ocular motor apraxia

### ***Report included:***

Matthew is a contented eight-month-old boy with Joubert's Syndrome. The Syndrome has impacted on his vision and his general development. He attended today with his Mum. He was alert and co-operative. Matthew was referred to the clinic by his paediatrician and is currently under the care of ophthalmology and community paediatrics. He has had glasses for two months and his mother reports that he is now more visually responsive. The glasses are a good fit and he wears them well. Matthew's mother is interested in gaining as much information as she can about what he can see and how she can help encourage his visual development.

### **Summary of findings:**

Matthew is very long-sighted and needs to be wearing glasses all the time. We have **increased his glasses to full strength** given his visual problems and his excellent compliance with spectacles. With his glasses on his high contrast vision is within normal limits for his age, but this does not mean his vision is 'normal'. **Matthew has a significant visual impairment due to his eye movement difficulty and this needs to be considered by his carers and other therapists.**

As is seen in Joubert's Syndrome, Matthew has a problem with eye movements and generalised low muscle tone affecting e.g. head control. It is difficult for him to make his eyes move voluntarily from side to side and he thrusts his head in the desired direction to force his eyes to move that way too. Matthew is also able to turn his eyes in to view things close to him and he can move his eyes up and down in a normal way to view things moving vertically.

When presenting objects and toys to Matthew it will be easier for him to see and respond to them if they are placed straight ahead of him. Objects may be moved up and down in this position and he should be able to follow them. Fast moving objects may be more difficult for him to track so the speed at which objects are moved may need to be considered.

Today Matthew failed to respond to objects presented away from a straight ahead position. This may be attributed to his eye movement difficulties but may also be a complication of the retinal dysplasia (abnormal development of the retina at the back of the eye) that is a part of Joubert's Syndrome. **This further underlines the importance of presenting objects, toys, spoons and bottles in a straight ahead position and giving Matthew time to "find" the object with his eyes.** He may find it easier to locate objects or toys with his eyes if he is also holding or touching them. In this way his other senses may be useful to help direct his eyes.

At eight months old Matthew's visual system is still developing and with time he may use the vision that he has much better. He will need regular visual assessment.

### Technical details

Current prescription (good condition, good fit):

R +4.50/-1.00x180 L +5.50/-2.00x180

Refractive error (1% cyclopentolate HCl):

R +8.00/-2.00x180 L +9.00/-3.00x180

Ocular posture: well controlled esophoria with glasses

Eye movements: **ocular motor apraxia**

Uses head thrusts to initiate lateral saccades

Fails to follow laterally but vertical movements and convergence normal

Stereopsis: Gross stereopsis present

Visual acuity (Cardiff acuity test @ 50cm) with glasses: Binocular 6/48

Accommodation (dynamic retinoscopy): grossly normal with full distance Rx

Gross confrontation: Peripheral restriction/neglect (needs further investigation in future)

Seen by: AN Other, \_\_\_\_\_ Senior Optometrist

*Cc: Parents, General Practitioner, Paediatrician, Ophthalmologist, Health visitor, Physiotherapist, Occupational and Speech & language Therapists*