Title: A Diagnostic Dilemma: Suspected Retinoblastoma in a Child from Sierra Leone

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A 3-year-old girl, previously treated in Sierra Leone for Retinoblastoma (RB), was seen at the Judith Kingston Retinoblastoma Unit, London, United Kingdom for a second opinion. She represented a diagnostic dilemma, as it was unclear whether her condition represented RB or whether she had pseudoretinoblastoma. We present this case to highlight the benefits of effective cross-border collaboration.

Based on clinical examination and CT imaging in Sierra Leone, which revealed a calcified intraocular mass in her left eye, she was initially diagnosed as having unilateral RB. Six cycles of systemic chemotherapy (vincristine, carboplatin and etoposide) were completed in Sierra Leone and enucleation was then recommended, which her family declined and a further opinion from London was sought.

Examination under anaesthesia in London revealed left cataract with 360-degree posterior synechiae and no discernible fundal view: hence a diagnostic dilemma. Right eye examination, including fluorescein angiography, was normal. The fluorescein angiogram was essential to exclude recognised pseudoretinoblastomas (eg bilateral Coats disease and asymmetric Familial Exudative VitreoRetinopathy). Ultrasound of the left eye demonstrated a thickened, possibly calcified, total retinal detachment (Figure 1). MRI imaging, reviewed in London and Amsterdam, demonstrated areas of T2 hypointense tissue within the retinal detachment without contrast enhancement and areas of T2 hypointensity/T1 contrast-enhancement mismatch[1] (Figure 2), indicating contrast leakage from the detached retina without a corresponding low-signal intensity mass on the T2 weighted images. Differential diagnosis included regressed RB and Coats Disease. Follow up with repeat MRI scan, reviewed again in London and Amsterdam, was planned. This showed no change after six months. Following multiple discussions with her family and between the multidisciplinary teams treating her across centres, enucleation is now planned, given the risks to her life of her condition potentially being RB outweighing the benefits of keeping the eye.

Despite collaboration between the treating teams in Sierra Leone and London, access to medical records and imaging results from Sierra Leone posed a significant challenge. For example, although a CT was done before treatment was commenced, the images were no longer available, as the family took the images away from the treating hospital and a copy of

the results were not kept in the child's medical notes. In London, we relied on the parental description of the CT report showing a calcified intraocular mass. A large proportion of patients referred to our unit (35%) do not have retinoblastoma and are frequently calcified.[2, 3]

The majority of retinoblastoma cases worldwide occur in low- and middle-income countries where outcomes are worse than in high-income countries.[4] Closer collaboration between RB centres, for example through the Global RB study and the RB-net links network, should help improve this disparity. [5] This will allow patients to have good quality care close to home and avoid long-distance travel seeking second opinions. If families do seek opinions in other countries it is vital to know previous examination findings and treatments. This case represents a practical example where both centres helped in the final care with the further opinion of a third centre.

References

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Figure 1: Ultrasound of left eye
Figure 2: MRI of the left eye.
Figure 2A and 2C are axial fat-suppressed T2 weighted images and Figure 2B and 2D are axial contrast-enhanced T1 weighted images with fat-suppression. The arrows point towards focal contrast enhancement in an area without a corresponding T2 low signal intensity mass.