Clinical Patient Management System and ERN-EYE: Solving rare eye diseases complex cases

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Summary
The Clinical Patient Management System (CPMS) is a secure web-based application created to facilitate the information exchange between experts of the European Reference Networks for Rare Eye Diseases (ERN-EYE). The Common system created by the European Commission was secondly adapted for ERN-EYE thanks to a specific dataset for ophthalmological examination. The aim of this presentation is to show how the CPMS helps to solve unclear clinical cases in three examples.

Case circuit within the CPMS (Figure 1)
To get help with managing a specific patient, the clinician has to upload the case using a specifically developed dataset available on the web application that was worked out by the experts in the field. By enrolling a patient on the application, the physician opens a new panel to share the case and is therefore designated as a panel lead. A panel composed of different experts can be invited and each expert gives an opinion on the case based on experts consensus. The application generates an outcome document which constitutes a recommendation for the treating physician.

Three examples of concrete cases solved using the CPMS

1. Using CPMS through comments
- 15-year-old female with bilateral papilledema and ocular pain
- No visual loss
- No signs of intracranial hypertension (IH)
- Carrier of Leber’s hereditary optic neuropathy (LHON) mutation
- No signs of acute stage of LHON
- Consultation request: possible explanation for this papilledema?

2. Using CPMS through contributions
- 2-month-old female with nystagmus and Peters anomaly (anterior segment dysgenesis) in both eyes
- Multisystemic malformations
- Consultation request: recommendations for treatment?

3. Using CPMS through videoconference tool
- 5-year-old female with genetically confirmed Stickler syndrome and significant photophobia
- High myopia and fibrous tissue in vitreous body of the right eye
- Old total retinal detachment, secondary glaucoma and chronic exudates in the left eye
- Consultation request: laser preventive treatment for the right eye

Outcomes
- Papilledema has decreased.
- Visual acuity is stable.
- Patient is under idebenone therapy.
- Follow-up every 6 months.

Conclusion
Through these three examples the CPMS has shown an effectiveness to manage difficult cases affected by rare eye diseases that cause visual impairment in children and young adults. It is the only application in Europe that has the power of easily accessing experts all over Europe in the spirit of the leitmotiv “the expertise travels, not the patient.” Thanks to this system having an opinion on a complex or rare case abroad has been made accessible both for the patient and the physician.