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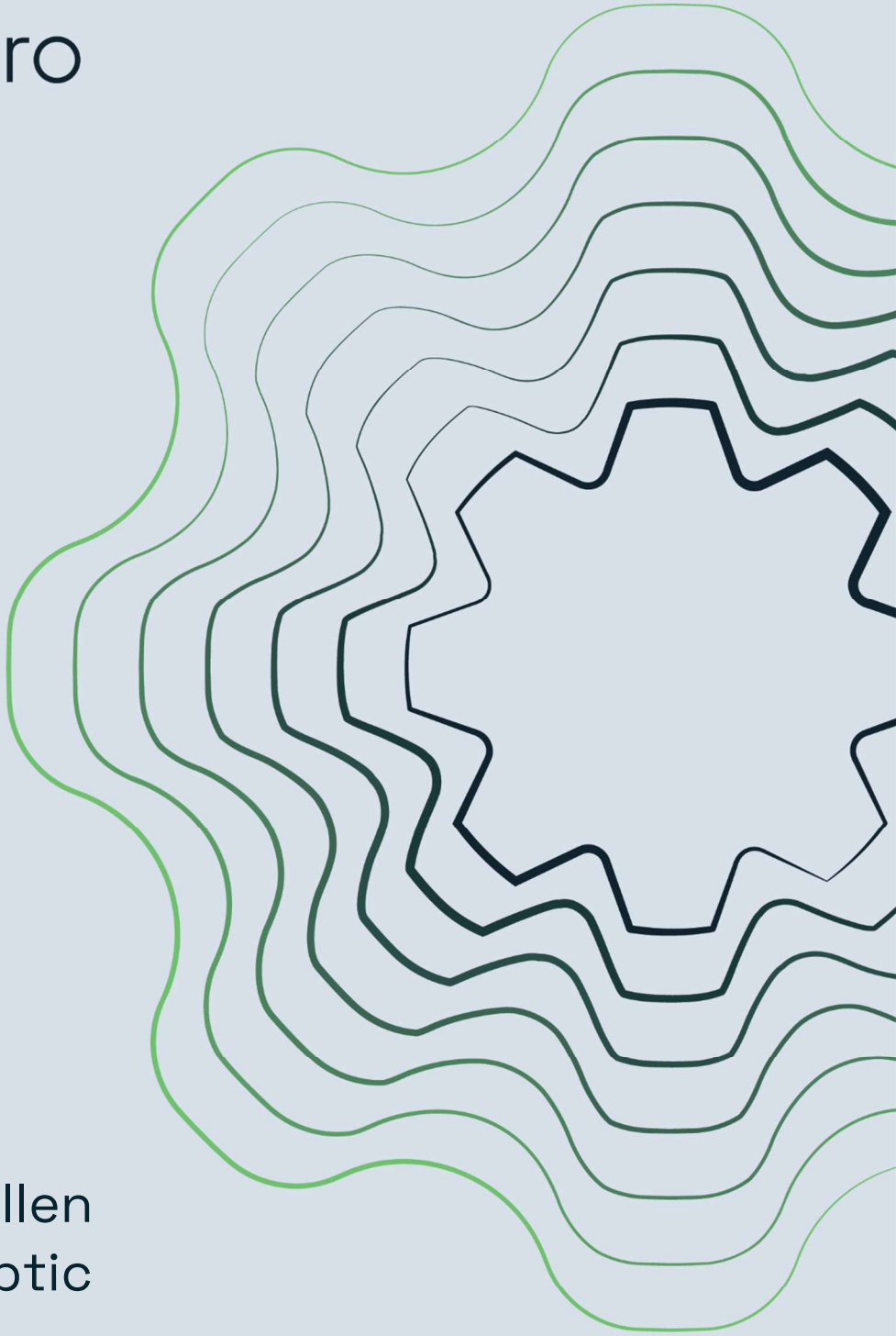

Approach to swollen appearance of optic discs in children

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Neurology and Developmental Medicine
for Children and Young People

Approach to Swollen Appearance of Optic Discs in Children

CYP Neuro Podcast episode1 hosted by Dr Ram Kumar in conversation with Dr Anil Israni

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Introduction

RK: Welcome, my name is Dr Ram Kumar. I'm a consultant paediatric neurologist and founding partner of CYP Neuro, which is an initiative to provide high-quality clinical and educational services in paediatric neurology. Today I'll be speaking to Dr Anil Israni, who is a consultant paediatric neurologist, friend, and fellow founding partner of CYP Neuro.

We're both experienced paediatric neurologists able to bring our combined wisdom and experience over many years to others such as yourselves listening, who would like to improve your knowledge and practice and learn from where we could have done things better.

In this first episode, we'll be talking about a common reason for referral to paediatric neurologists and paediatricians: a child or young person with swollen appearance of the optic discs. This finding can cause concern for doctors, other clinicians, and families because we know there are benign and serious causes which can be associated with vision loss or life-threatening conditions. We know the serious causes need to be identified and treated quickly to avoid adverse outcomes such as blindness, but we also know most children do not have serious causes, so we don't want to subject them to unnecessary invasive investigations like lumbar puncture or general anaesthetic.

Let's get some practical insights and clinical approach to this problem from Dr Israni, who's well experienced with dealing with this common scenario. We'll approach this from our perspective as non-ophthalmologists, covering some of the common pitfalls and how to avoid them. So thank you, Dr Israni, for joining me today. Can you remind us what the optic discs

actually are and why we've chosen the topic name "Approach to Swollen Appearance of Optic Discs" instead of just "Approach to Swollen Optic Discs" or "Approach to Papilloedema"?

Understanding Optic Discs

AI: Thank you for inviting me, Dr Kumar. Essentially, the optic discs are the visible portion of the optic nerve that are visible in the back of the eyeball and are seen through the pupil using an ophthalmoscope or sometimes special imaging. The optic discs contain the entry or exit of the optic nerve axons between the brain and the retina. It is thus a window into the brain for neurologists. It also contains blood vessels. It is in the shape of a disc, hence the name optic disc.

The optic disc can become oedematous or swollen in neurological conditions, eye diseases, as well as some systemic conditions. However, a swollen appearance of optic disc does not necessarily mean optic disc oedema. There are many causes and entities that can give rise to such appearance. Some are of ophthalmological or ocular origin, others are neurological or systemic. It is therefore important for paediatricians and paediatric neurologists to have familiarity with these ophthalmic and neuro-systemic conditions which can give rise to swollen appearance of optic disc. The term papilloedema is reserved for true swollen optic disc or discs (unilateral or bilateral) due to raised intracranial pressure.

Terminology and Clinical Presentation

RK: Okay, so that's swollen discs or swollen optic disc, but I also hear about blurred disc margins as well. Is that different or the same as swollen appearance of the optic discs or papilloedema?

AI: It essentially refers to the same problem, and we come across the term blurred disc margins being used interchangeably with swollen discs or papilloedema or disc oedema or sometimes

papillitis. The margins of the optic disc become blurred when the optic disc appears swollen. But it is important to be precise because these terms are relatively specific.

Serious Causes of Swollen Optic Discs

RK: That's an important point you raised there about being specific, and this issue about communication and miscommunication between colleagues about swollen optic disc appearance is important. We'll discuss ways how we can improve this communication as well. So what are the other serious entities or conditions that can result in appearance of swollen optic discs?

AI: The true oedema of the optic disc due to raised intracranial pressure and its underlying causes, or related to causes other than raised intracranial pressure (sometimes referred to as papillitis), can both give rise to swollen appearance of the optic disc.

The causes of swelling of the optic disc other than raised intracranial pressure can have a myriad of causes including:

- Inflammatory and demyelinating conditions, for example, optic neuritis such as anti-MOG or anti-Aquaporin-4 related demyelination
- Infectious causes like Lyme disease
- Infiltrative causes like leukaemic infiltration of the optic nerve
- Ischaemia and hypertension, which are common causes of ischaemic optic neuropathy in adults
- Toxic, drug-induced and nutritional causes
- Compression of the optic nerve due to intra-orbital masses
- Genetic conditions, for example, Leber's hereditary optic neuropathy

These entities can also cause a more posterior retrobulbar optic neuritis or dysfunction, and these might not show any obvious fundus or disc abnormalities on the ophthalmoscope, but these may be visible on special imaging.

Clinical Assessment

RK: Logically, given that the majority of cases with concern of optic disc swelling do not actually have true papilloedema, what's the first step in the assessment to establish if the swollen appearance is true or due to a mimic? How do we go about this?

AI: For a clinical neurologist, the clinical context is as much or probably more relevant than just the appearance of optic disc. Concluding raised intracranial pressure to be present based merely on swollen appearance of optic disc is a potential pitfall and we need to go back to the basics of obtaining relevant clinical history with mapping of clinical onset and the trajectory.

We need to establish presence or absence of symptoms related to raised intracranial pressure, particularly:

- Recent onset of progressive headache
- Headache associated with Valsalva manoeuvres
- Worsening on sneezing, coughing, lying down or bending forwards
- Presence of other red flags related to headache symptomatology like:
  - Nocturnal waking or morning headache
  - Neck pain or back pain
  - Vomiting
  - Pulsatile tinnitus
  - Visual blurring
  - Eye pain
  - Transient visual obscurations
  - Double vision

We should also look for any systemic risk factors, medication history, and family history.

The relevant points on general physical examination would be:

- Body weight and BMI
- Blood pressure
- Head size and shape
- Presence of any craniofacial dysmorphism
- Suture regions for any cranial deficit
- Abducens nerve palsy
- Other features of raised intracranial pressure
- Any focal neurological deficits
- Any signs of meningeal irritation

In an office setting, a paediatric neurologist can perform some basic ophthalmological assessment:
- Visual acuity using Snellen chart
- Colour vision using Ishihara charts
- Eye movements for any paralytic squint
- Gross visual field assessment using confrontation method
- Fundus examination using an ophthalmoscope

There are limitations to what information can be obtained from a toddler or young child, both with regards to clinical history and clinical examination due to issues with cooperation.

Role of the Ophthalmologist

RK: We rely a lot on the ophthalmologist's eye assessment and often they've seen the child before us. What do you think is the ophthalmologist's role in assessing these children?

AI: An experienced ophthalmologist, especially a neuro-ophthalmologist, can based on skill and experience reliably differentiate between various entities giving rise to swollen appearance of optic discs. They look for several key features:

- Spontaneous venous pulsation
- Blood vessel margin patterns
- Retinal peripapillary haemorrhages
- Colour vision changes
- Eye movement abnormalities

In optic neuritis, colour vision may be affected early and the eye movements tend to be painful, whereas loss of visual acuity is not typical early in papilloedema unless there is macular oedema, and eye pain is often absent. There may be eye movement abnormalities in either condition due to the false localising feature of sixth nerve palsy or internuclear ophthalmoplegia.

Ophthalmologists tend to dilate the pupils with mydriatics for examination, although there's no difficulty in principle for such to be done by non-ophthalmologists. However, eye examination in children can be challenging even for experienced ophthalmologists. Not all ophthalmologists will be aware of the range of differential diagnoses that we discussed earlier and the discriminative value of clinical history and examination features.

Ancillary Investigations

RK: Okay, so you've mentioned some ancillary investigations. We want to get to the underlying cause efficiently and in a timely way without unnecessarily subjecting the majority of children who we already know may not have a serious underlying cause to invasive tests or general anaesthetic. What would be your approach in terms of the ancillary investigations?

AI: Much will depend on local availability and expertise. Several key investigations can be helpful:

<u>Ultrasound B-scan</u>

- A useful, simple, non-invasive, outpatient-based test

- Performed by radiologists and in some centres by ophthalmologists
- Can help identify optic nerve head drusen, especially if calcified
- Can show dilatation of the optic nerve sheath and raised optic nerve heads
- Shows as a hypoechoic nodule beneath the optic nerve head at low gain level
- In acquired disc oedema, demonstrates a "crescent sign" within the optic nerve sheath
- This crescent sign is produced by increased cerebrospinal fluid transmitted along the subdural space

Optical Coherence Tomography (OCT)

- Can help differentiate papilloedema from optic nerve head drusen
- Useful in treatment monitoring
- Measures retinal nerve fibre layer thickness
- Correlates with CNS nerve axon integrity
- Retinal nerve fibre layer thickness is usually high in papilloedema
- Shows characteristic shadows and lumps in drusen
- Can be used to monitor treatment response

Fluorescein Angiography

- Involves intravenous dye injection
- Looks for dye leakage in true optic disc swelling
- Shows hyperfluorescence and peripapillary leakage in papilloedema
- No leakage in congenital disc elevation
- Invasive test requiring child cooperation

Imaging Studies

RK: What is the next set of investigations that we should use to narrow down the underlying diagnosis, particularly if we think it is true disc swelling?

AI: The imaging modality of choice is MRI of the brain with MR Venography (MRV). This helps identify:

- Mass lesions
- Hydrocephalus
- Cerebral oedema
- Cerebral venous sinus thrombosis

MRI of the orbits, particularly T1 with fat suppression, may also be relevant if there is:

- Vision impairment
- Eye movement restriction
- Eye pain

MRI of the brain is also relevant to look at other causes of papillitis disc swelling without raised intracranial pressure. The majority of children with disc swelling will require an MRI of the brain even when there are no signs of raised intracranial pressure since alternative causes of papillitis often have CNS involvement.

CT head is acceptable in emergency situations, especially in:

- Head injury due to trauma
- Suspicious signs of progressive raised intracranial pressure
- Meningitis

However, CT head has limited sensitivity for posterior fossa lesions, which are relatively common in children compared to adults. Idiopathic intracranial hypertension (IIH) will also not be identified or excluded on a CT scan.

<u>Ultrasound B-scan</u>

- A useful, simple, non-invasive, outpatient-based test
- Performed by radiologists and in some centres by ophthalmologists
- Can help identify optic nerve head drusen, especially if calcified

- Can show dilatation of the optic nerve sheath and raised optic nerve heads
- Shows as a hyperechoic nodule beneath the optic nerve head at low gain level (characteristic of drusen)
- In acquired disc oedema, demonstrates a "crescent sign" within the optic nerve sheath
- This crescent sign is produced by increased cerebrospinal fluid transmitted along the subdural space

Diagnostic Criteria for IIH

RK: In our usual practice as paediatric neurologists, we often receive referrals for children who have only had an MRI brain scan, sometimes requested by the ophthalmologist and they've usually been undertaken without contrast or the MRV sequence. I'm going to assume here that a significant mass lesion or anatomical finding like hydrocephalus will be detected on the standard MRI brain scan, but as we discussed earlier, the majority of children will not have abnormalities. What clues are there for idiopathic intracranial hypertension being visible on an MRI brain before we get to having to do an invasive lumbar puncture study to confirm or identify raised pressure?

AI: Fundamentally, there are diagnostic criteria for IIH, which are based on radiological as well as lumbar-puncture-based CSF indices.

<u>Radiological Prerequisites:</u>
- Absence of mass lesion, hydrocephalus, or structural abnormality
- Absence of cerebral venous sinus thrombosis on MRI with MRV

<u>CSF Prerequisites:</u>
- Normal CSF constitution (excluding CNS inflammation like meningitis, encephalitis)
- Objective demonstration of raised intracranial pressure through CSF opening pressure measurement

MRI Findings Suggestive of IIH:

These findings are not universally present nor specific to intracranial pressure:

- Tortuosity of the optic nerves
- Dilated perioptic nerve sheaths
- Empty sella
- Posterior scleral flattening
- Protrusion of the optic nerve head into the orbit
- Transverse sinus stenosis, particularly distal (requires MRV)

Additional Investigations for Non-IIH Cases

RK: What other assessments should we undertake if we suspect that this is not papilloedema due to raised intracranial pressure, but rather a papillitis of unknown cause?

AI: With CSF manometry, any pressure more than 28 centimetres of water is definitely raised intracranial pressure and more than 25 centimetres of water is probably raised intracranial pressure.

For other causes of papillitis, investigations should be planned based on the clinical picture:

Basic Investigations:

- Full blood count
- Urea and electrolytes
- Blood lactate
- Autoantibody profile

Specific Tests:

- Anti-MOG or anti-Aquaporin-4 antibodies (for suspected demyelination)
- CSF for paired oligoclonal bands
- Vitamin B12 levels (for nutritional causes)

- Visual Evoked Potentials (VEP) for retrobulbar optic neuritis
- Electroretinogram (ERG) for retinal problems
- Genetic testing (e.g., for Leber's hereditary optic neuropathy)

Best Practice Recommendations

RK: Do you have any further tips on how to improve practice, getting to the diagnosis and treatment fast, but not over-investigating the majority of children who do not need invasive investigations when there's a swollen appearance of the optic discs?

AI: My experience suggests several key approaches:

1. Direct Communication:
   - Talk to the ophthalmologist directly
   - See children jointly in clinic when possible
   - Review investigations together including OCTs and retinal photographs
   - Correlate findings with symptoms and signs

2. Imaging Review:
   - Review neuroimaging with the neuroradiologist
   - Don't rely solely on written reports

3. Standardised Approach:
   - Use standardised grading systems (e.g., Frisén grading system from one to five)
   - Learn visual examination techniques from ophthalmologists
   - Utilise new technology like handheld non-mydriatic retinal cameras
   - Consider point-of-care wireless portable ultrasound

Key Takeaway Messages

RK: What are your takeaway lessons for the paediatrician or practising paediatric neurologist evaluating a child with a swollen appearance of the optic discs?

AI: The key takeaway lessons are:

1. There's no substitute for meticulous clinical history and examination
2. Majority of asymptomatic children will not have true optic disc oedema or papilloedema
3. Consider serious neuro-systemic and eye conditions beyond raised ICP
4. A normal MRI brain or normal lumbar pressure should not be the end of assessment
5. Improving communication between ophthalmologists, neurologists and neuroradiologists is essential
6. An MDT-based approach is crucial, especially in grey areas
7. There is much to learn from each specialty involved

RK: Thank you very much Dr Israni for joining me today in discussing this important topic. This is just one of a series of podcasts and we'll share our experience with further childhood neurological conditions and scenarios in future podcasts. We'll be returning to the theme of raised intracranial pressure and papilloedema by interviewing other colleagues in ophthalmology amongst other relevant specialists. Listeners can send in their questions and provide their comments and feedback via our LinkedIn pages or through the CYP Neuro website. On the website, we'll also share a transcript of this podcast episode and further references on the CYP Neuro Academy web pages. Thank you very much once again.

AI: Thank you.