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From "just reflux" to lissencephaly: An infant's diagnostic journey

Diagnostic overshadowing in early infancy can delay the identification of rare congenital neurological disorders. This case highlights the clinical challenge of distinguishing atypical paroxysmal events from benign infant phenomena, the critical role of neuroimaging in unexplained neurological signs, and the multidisciplinary management required for complex cortical malformations.

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Key points

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1. Type 1 lissencephaly is a cortical malformation caused by abnormal neuronal migration, diagnosed definitively via brain MRI.
2. Infants with lissencephaly carry a high risk of seizures and neuro-developmental impairment, requiring close monitoring and multidisciplinary care.
3. Proactive parental engagement, genetic counselling and early developmental support are vital components of comprehensive patient management.

Introduction

Gastro-oesophageal reflux is a common occurrence in early infancy, frequently presenting with unsettled behaviour, arching and vomiting. However, recurrent paroxysmal episodes accompanied by atypical features such as gaze abnormalities or limb extension warrant careful neurological evaluation.[1] Lissencephaly is a severe malformation of cortical development characterised by the absence or reduction of cerebral convolutions (gyri), resulting in a smooth brain surface.[2] Prompt recognition is essential for early risk stratification, seizure management and parental counselling.

Case presentation

A two-month-old infant presented to the paediatric department with recurrent episodes of repetitive back-arching. The infant was born at 40^{+0} weeks' gestation via spontaneous vaginal delivery to non-consanguineous parents. Immediate postnatal adaptation was complicated by respiratory distress requiring a 36-hour admission to the NICU with supplemental oxygen and intravenous antibiotics. Prior to this evaluation, recurrent back-arching episodes had been observed by healthcare professionals and attributed to gastro-oesophageal reflux. On clinical examination, the infant demonstrated repetitive back-arching accompanied by staring eyes, extension of the left arm and left eye exotropia. The remainder of the neurological examination was unremarkable, and no obvious dysmorphic features were noted. A comprehensive neuro-metabolic workup yielded normal routine blood investigations. Magnetic resonance imaging (MRI) of the brain subsequently revealed Type 1 lissencephaly, characterised by a thickened cortex and deficient neuronal migration.[1,2] Given this neurodevelopmental finding and parental concern regarding observed paroxysmal events, an initial electroencephalogram (EEG) was performed, which showed no epileptiform activity.[2] Following multidisciplinary consultations with tertiary paediatric neurology centres, initiation of antiepileptic therapy was recommended due to the high risk of seizures associated with lissencephaly. Further outpatient ambulatory EEG and genetic testing were also advised. A subsequent repeat EEG demonstrated electroclinical features of epileptic seizures. Consequently, a second antiepileptic agent was introduced to optimise seizure control and reduce overall neurological burden.

Discussion

Lissencephaly is a spectrum of rare brain malformations resulting from arrested or impaired neuronal migration between the 12th and 24th weeks of gestation.[2,3] It is broadly classified into Type 1 (classic lissencephaly, featuring a four-layered cortex) and Type 2 (cobblestone lissencephaly, characterised by a disorganised, pebbled cerebral surface).[3] Postnatal diagnosis relies primarily on neuroimaging, as the clinical presentation encompasses non-specific neurological findings.[3,4] Advances in molecular genetics have identified numerous LIS-associated genes, enabling genotype-phenotype correlations that aid in predicting structural cortical patterns and clinical

severity.[1,4] The clinical manifestations of lissencephaly include global developmental delay, severe psychomotor impairment, mental disability, feeding difficulties, hypotonia progressing to spasticity and intractable epilepsy.[2,3] Management is primarily supportive and multidisciplinary, focusing on nutritional support, respiratory care and antiepileptic therapy.[2,3] The prognosis remains guarded, with high morbidity and a significant mortality rate estimated at approximately 50% by 10 years of age, frequently driven by aspiration pneumonia, respiratory infections and status epilepticus.[3,4] This case underscores the importance of maintaining a broad differential diagnosis when evaluating recurrent or atypical symptoms in infants, particularly when initial conservative management for suspected reflux fails.[1] Early recognition through neuroimaging and clinical genetics facilitates proactive surveillance, empowers families through informed counselling and ensures timely intervention for emerging complications.[1,4]

Parental consent

Written informed consent was obtained from the child's parents for the publication of this case history and associated clinical details.

References

1. **Dobyns WB, et al.** Lissencephaly: expanded imaging and clinical classification. Am J Med Genet A 2018;176:2095-103.
2. **Kattuoa MI, Das JM.** Lissencephaly. Stat Pearls. Online at: www.ncbi.nlm.nih.gov/books/NBK560766
3. **Barkovich AJ, et al.** A developmental and genetic classification for malformations of cortical development. Neurology 2005;65:1873-87.
4. **Kidokoro H, et al.** Clinical and neuroimaging features of lissencephaly: A retrospective study. Brain Dev 2016;38:54-62.

22-24 OCTOBER

Neo Care 2026

The World Congress on Pediatrics, Child Health and Neonatal Care will take place in Orlando, USA, with an option to join virtually from anywhere in the world. Organised by the Iris Scientific Group, Neo Care 2026 aims to unite pediatricians, neonatologists, child health specialists, nurses, researchers and healthcare professionals to share knowledge, exchange ideas and discuss the latest advancements in child and neonatal care.

Venue: Hilton Garden Inn Lake Buena Vista/Orlando, USA

Contact: pediatrics@irissciencegroup.com

28-31 OCTOBER

Congress of the European Paediatric Academic Societies

CEPAS 2026 is the new biennial meeting of the European Academy of Paediatrics (EAP) and the European Society for Paediatric Research (ESPR). All paediatric subspecialties, spanning from neonatal care to adolescent medicine, come together to advance child health through cutting-edge research, education and collaboration. For over 15 years, EAP and ESPR have been leading the way in paediatric research, education and collaboration. The collaboration between ESPR and the Global Foundation for the Care of Newborn Infants enhances the integration of patient and family-centred perspectives, reflecting CEPAS' commitment to interdisciplinary dialogue.

Venue: Lyon Convention Centre, France

Contact: www.cepas.org

1-4 NOVEMBER

NETNEP 2026: 10th International Nurse Education Conference

The NETNEP conference series facilitates the global exchange of knowledge and expertise in nursing, midwifery and healthcare education. The 10th conference will explore how educational innovations enhance student learning experiences, support the professional development of qualified practitioners, and improve health outcomes and safety for individuals and communities worldwide.

Venue: Edinburgh, UK

Contact: conferenceinfo@elsevier.com
www.elsevier.com/en-gb/events/conferences/all/international-nurse-education-conference

10-12 NOVEMBER

Neonatal World Conference 2026

The theme is Frontiers in Neonatology: Beyond Survival to Success. The Neonatal World Conference is a global platform that brings together leading experts in the field of neonatal care to enhance the health outcomes of newborns worldwide collaboratively. This international event will bring together neonatologists, paediatricians, researchers and healthcare professionals from around the world to share experiences, explore innovative technological advancements and discuss the ongoing challenges in neonatal and perinatal medicine.

Venue: Village Hotel Changi, Singapore

Contact: neonatal@precisionglobalconferences.com
www.neonatalconference.precisionglobalcon.com/about

13-14 NOVEMBER

NeoFOCUS UK – Introduction to Neonatal Haemodynamics

Day one is a lectures-based course for beginners and advanced learners in neonatal echocardiography. Day two is a hands-on workshop for beginners to understand neonatal functional echocardiography.

Venue: The Lowry Hotel, Chapel Wharf, Salford

Contact: neofocusUK@gmail.com
www.neofocus-uk.com

19-20 NOVEMBER

Neonatal Palliative and End of Life Care Simulation Course

In collaboration with the Northwest Neonatal ODN and Manchester University NHS Trust, this course has been developed to allow an immersive experience of neonatal palliative and end-of-life care lectures, discussions and small group simulations. Topics include: death in the delivery room; decision making at the cusps of viability, antenatal counselling for terminal conditions, application of parallel planning and advanced care planning concepts and bereavement clinic follow-up.

Venue: Whiston Simulation Suite, Education Centre, Whiston Hospital, Prescot

Contact: nest.courses@mft.nhs.uk or leanne.beaumont@mft.nhs.uk

19-22 NOVEMBER

11th International Neonatology Association Conference

This year, the conference will explore the central theme Effects of Early Interventions for a lifetime. With this focus, the conference will bring together renowned international speakers and leading experts to examine long-term outcomes and the latest solutions in neonatal care.
INAC 2026 will feature a dynamic programme, including pre-conference workshops led by world-class experts, engaging scientific sessions addressing the most pressing topics in neonatology, and an exceptional networking opportunity for neonatologists, perinatologists, paediatricians, nurses, nurse practitioners and respiratory therapists from around the world.

Venue: Novotel Amsterdam City, the Netherlands

Contact: secretariat@worldneonatology.com
www.worldneonatology.com/2026

23-24 NOVEMBER

UK National Neonatal Transport Group Conference 2026

Organised by the Northern Neonatal Transport Service, the programme will focus on practical learning, innovation and real-world application, including: Intrauterine transfer – experience, challenges and implementation; simulation in transport training – using it to adapt and evolve practice, with learning fed back into the neonatal unit environment; family integrated care – innovation and best practice within the transport setting.

Venue: Gateshead, Newcastle

Contact: www.cfsevents.co.uk

23-26 NOVEMBER

Neonatal Update 2026

Neonatal Update is an established annual event produced by Imperial College London. It has origins in a meeting established over 40 years ago and is renowned for its innovative programme incorporating contributions from a wide range of world-leading scientists and clinical researchers. The aim is to introduce delegates to areas of science not necessarily met in their day-to-day activities, to widen horizons and to foster new collaborations.

The 2026 meeting will be a four day in-person event, to allow the best possible opportunity for knowledge sharing, discussion and networking with the global neonatology peer group in the same space.

Venue: BMA House, London

Contact: www.imperial.ac.uk/school-public-health/neonatal-update/neonatal-update-2026

27-28 NOVEMBER

Efficient Use of Pocus in NICU

A two-day course for neonatal and paediatric consultants and trainees, specialty doctors, ANNPs and other medical doctors interested in learning and improving their lung ultrasound and other POCUS skills. Day one includes basics and physiology of lung USS, normal and abnormal lung ultrasound findings, POCUS for vascular catheters, when vascular catheters go wrong, PDA haemodynamic assessment, looking at the heart – the basics, ultrasound of a collapsing baby and endotracheal tube position adjustment.Day two will include practical hands-on sessions, interactive video sessions and MCQ evaluation. Hybrid available on day one.

Venue: Education Centre, St Peter's Hospital, Surrey

Contact: https://l1nq.com/7dpbrxl

18-19 JANUARY 2027

UK Preterm Birth Conference 2027

The conference brings together internationally recognised leaders in maternal, fetal and neonatal health for two days of scientific exchange, multi-disciplinary collaboration and innovation-driven discussion focused on reducing the global burden of preterm birth.

Hosted in East London at Queen Mary University of London, the conference will feature 15 keynote lectures, four plenary talks, scientific discussions, presentations and training workshops, uniting experts from medicine, engineering, public health, translational science, digital health, policy and industry. The programme explores major themes shaping the future of preterm birth prevention and care: Prediction and diagnostics; prevention strategies; neonatal outcomes; health and policy; AI, digital twins and virtual healthcare systems; and translational research.

Venue: Peoples Palace, Queen Mary University of London, Tower Hamlets

Contact: t.t.chowdhury@qmul.ac.uk
https://pretermconference.com

29 JANUARY

NWNODN Annual Conference 2027

For those looking to expand their knowledge, share ideas, showcase work, or celebrate excellence in neonatal care, this conference offers the ideal platform to connect, inspire and grow. It will bring together international and national faculty, sharing the latest evidence, innovations and developments in neonatal care.

Key topics include: Recent advances in surfactant administration and management of RDS; beyond hypothermia – next generation neuroprotective agents for HIE; automated neonatal ventilation using AI – Myth or reality; and lived parent experience.

Venue: The Edge, Riveredge, Wigan

Contact: www.neonatalnetwork.co.uk/nwnodn/event/nwnodn-annual-conference-2027

21-22 JUNE

REaSoN Neonatal Meeting 2027

REaSoN has evolved over a 30+ years as an educationally rewarding annual conference for doctors, nurses, ANNPs, nurse educators, HPPPs or AHPs, psychologists and pharmacists with a shared interest in all aspects of neonatology. The meeting provides dedicated sessions that cover current research evidence, quality improvement, nursing hot topics, ethics and frontier science. Leading UK and International speakers are attracted, and lively debate encouraged.

This hybrid meeting will allow delegates to view educational content, partake in Q&A and live polling sessions with speakers, liaise with other delegates in networking groups, speak to exhibitors, view poster presentations and participate in networking.

Venue: Chesford Grange Hotel, Kenilworth and OnAIR conference platform

Contact: www.reasonmeeting.co.uk

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