Department of Health & Social Care

From [redacted]
Parliamentary Under-Secretary of State for Public Health and Prevention

39 Victoria Street
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Our ref: [redacted]

HM Coroner Alan Anthony Wilson
HM Coroner Blackpool and Fylde
PO Box 1066
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By email: [redacted]

2 October 2024

Dear Mr Wilson,

Thank you for the Regulation 28 report of 12 July sent to the Secretary of State for the Department of Health and Social Care about the death of Ryleigh Hillcoat-Bee**.** I am replying as the Minister with responsibility for rare diseases.

Firstly, I would like to say how saddened I was to read of the circumstances of Ryleigh's death, and I offer my sincere condolences to their family and loved ones. The circumstances your report describes are very concerning and I am grateful to you for bringing these matters to my attention.

The report raises concerns over:

- the lack of awareness of rhabdomyolysis and LIPIN-1 deficiency, a rare condition, amongst paediatricians in general paediatric departments
- the likelihood that previous cases of rhabdomyolysis in young children have been missed
- the limited amount of available guidance
- in the event other young children attend a general paediatric department for reasons connected to rhabdomyolysis, the condition will go unrecognised and with fatal consequences.

In preparing this response, my officials have made enquiries with NHS England and NICE to ensure we adequately address your concerns.

I recognise that people living with rare diseases such as LIPIN-1 deficiency face numerous challenges and more can be done to prevent such deaths in the future. You might be aware that in England, we have now published three action plans setting out how we will address the four priorities of the UK Rare Diseases Framework (the Framework). To ensure delivery

and accountability, each action lists an owner, desired outcomes and how we will measure and report on progress. Of most relevance to the concerns raised is the priority around increasing awareness of rare diseases among healthcare professionals. The other three priorities are: helping patients get a final diagnosis faster, better coordination of care, and improving access to specialist care, treatment and drugs.

With over 7,000 rare diseases, it is not possible for healthcare professionals to receive comprehensive training on every condition. Therefore, it is important that they are aware of rare diseases more broadly and are alert to considering them. Actions undertaken to increase awareness of rare diseases among healthcare professionals include:

- developing an innovative digital educational resource by the Genomics Education Programme (GEP)
- determining how best to include rare diseases in UK health profession education and training frameworks
- extending the remit of the GEP to include non-genetic rare diseases
- publishing high-quality epidemiological and research papers to increase the understanding of rare diseases
- publishing and implementing specific strategies for increasing awareness of rare diseases in the nursing and midwifery, pharmacy and primary care workforce
- developing a genomics communication skills resource
- developing a specialist genomics workforce through the Genomics Training Academy (GTAC)

Officials engaged NICE with respect to your concern around a lack of guidance on rare diseases. NICE state that they do not usually produce guidance on the management of rare diseases as there is usually limited high quality evidence available on which to base guidance on. Therefore, guidance for rare conditions is usually developed by groups of interested clinicians or specialist societies and by consensus. NICE advise that specialist guidance already exists for the conditions in question, and, with limited evidence, they would not be able to go beyond that existing guidance.

Additionally, NICE have advised that they consider the awareness of rare conditions is best addressed through education and dissemination activities. This could be done through NHSE, professional societies (e.g. Royal College of Paediatrics and Child Health (RCPCH)), specialist and other groups. Further, that the recognition and management of metabolic emergencies and rhabdomyolysis in Local guidance should cover the practicalities of investigation, management and referral, which will vary according to local practice.

The NHSE advise that more broadly, they recognise that rhabdomyolysis is a rare but potentially lethal condition, which according to the National Child Mortality Database, contributes to approximately four deaths in children each year. Many cases are associated with trauma and associated compartment syndromes, and it should be recognised that although contributory it may not be entirely causative. Very few will be associated with Lipin A deficiency.

Following on from the above, the NHSE confirm that they approached the RCPCH who are the professional body responsible for postgraduate education of paediatricians. NHSE inform us that RCHCP are also intending to respond to this report, although I recognise, they are not official recipients of this report. I will, thus lay out in detail the information provided

to NHSE by RCHCP. The information provided by RCPCH clarifies that in line with the GMC expectations of curricula and 'excellence by design', they do not have a list of specific conditions included. Their curriculum is an outcomes based one with high level generic learning outcomes which focus on professional and clinical behaviours and capabilities.

Rhabdomyolysis is a far from common diagnosis in children, and they would not expect many doctors to have had direct experience of managing it during their training. However, RCHCP would expect them to have some awareness of it and to understand that they may encounter many unusual presentations in children. The generic curriculum emphasises working with the multidisciplinary team and considering the full range of diagnoses and as outlined in the general paediatric specialty level syllabus, drawing on the expertise of specialists as appropriate.

Examples of the specialty level curriculum which talks about these capabilities are:

- Learning outcome 2: Effectively communicates with the multi-disciplinary team (MDT), children, young people and their families, where there is a range of differential diagnoses and management is uncertain
- Learning outcome 4: Considers the full range of differential diagnosis, treatment and management options available. Supervises junior staff and supports colleagues in the assessment and management of cases which are complex or nuanced.

These capabilities should build on medical student and foundation training which RCPHC would expect to cover clinical reasoning and the development of 'professional curiosity'. rhabdomyolysis is featured in the Intercollegiate General Surgery curriculum, and it would be expected for all surgeons to be aware of this condition. (https://www.gmc-uk.org/-/media/documents/General_Surgery_inc._Trauma_TIG__approved_Jul_17_.pdf_72509288.pdf). Rhabdomyolysis is also covered in the Faculty of Intensive Care Medicine curriculum and on numerous educational materials: https://www.ficm.ac.uk/index.php/documents/key-take-home-messages-21. In conclusion, much postgraduate paediatric education is focussed on recognition of the seriously unwell child and onward referral. It would not be reasonable or effective for all paediatricians to be completely up to date on diagnosis and management of rhabdomyolysis but it would be expected that they would recognise serious illness and involve the appropriate experts.

In relation to your concern that in the event other young children attend a general paediatric department for reasons connected to rhabdomyolysis, the condition will go unrecognised and with fatal consequences. NHS England’s Genomics Education Programme (GEP) are developing an innovative digital educational resource (GeNotes) - providing healthcare professionals with relevant and concise information to support patient management, linking to the NHS Genomic Test Directories, and signposting to extended learning opportunities. With their subject matter experts and paediatric working group, they can quickly develop resources relating to rhabdomyolysis for paediatricians in general paediatric departments. The GEP are also working with Royal Colleges and higher education providers to include rare diseases in education frameworks and curricula. It is crucial to provide future healthcare workforce with the knowledge and awareness of rare diseases to support patients and their families and prevent future deaths. In addition, patient organisations and advocacy groups

are also included in the development of education and training which enhances understanding of the challenges faced by the rare diseases community.

The update from NHSE on the GEP also recognises that serving the rare diseases community through raising awareness of genomics and educating and training the NHS workforce is a key strategic and operational priority for the programme. A senior representative from the GEP has been a member of the UK Rare Diseases Delivery Group since its inception in 2021. The main focus for the GEP in the Framework is priority two which is, increasing awareness of rare diseases among healthcare professionals.

Referring to the concern in particular around lack of awareness amongst paediatricians in general paediatric departments. The GEP is utilising frameworks and educator toolkits to deliver education and training and raise awareness of rare diseases to the wider workforce including nursing and midwifery, pharmacy and primary care. Their specialists in these areas can advise and steer on the resources required to meet the needs of each individual workforce area.

The GEP is also developing a three-tier communication skills education resource for the wider and specialist workforce. Tier one will cover core communication skills, tier two application of skills in practice and tier three highly specialist skills for the genomics workforce delivering the Genomic Medicine Service (GMS). These packages of learning are being developed with patients and the public with lived experience and a number of charities and support groups, including M4RD and the Personalised Care Institute (PCI). They will equip the specialist and wider workforce to have compassionate and patient-centred conversations, aligned to shared-decision making approaches."

With respect to the specific concerns raised in the report, where guidance and workforce awareness appear to be limited and further education and training is required, the GEP will contribute by working with the Department and in collaboration with Medics for Rare Diseases (M4RD) on a number of solutions aligned to some of the actions above.

I welcome the contributions made by delivery partners and look forward to supporting work to improve the lives of people living with rare conditions.

I hope this response is helpful. Thank you for bringing these concerns to my attention.

Yours sincerely,

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PARLIAMENTARY UNDER-SECRETARY OF STATE FOR PUBLIC HEALTH AND PREVENTION