



## Consultation response

### Antimicrobial resistance delivery plan

Response by Genetic Alliance UK, 25th February 2016

#### Introduction

1. Genetic Alliance UK is the national charity working to improve the lives of patients and families affected by all types of genetic conditions. We are an alliance of over 180 patient organisations. Our aim is to ensure that high quality services, information and support are provided to all who need them. Genetic Alliance UK actively supports research and innovation across the field of genetic medicine.
2. Rare Disease UK is a multi-stakeholder campaign run by Genetic Alliance UK, working towards the delivery and implementation of a national strategy for rare diseases in the UK. Rare Disease UK has actively supported collaboration between the four health ministers of the UK to sign up to the UK Strategy for Rare Diseases<sup>1</sup>.
3. There is much more that could and should be done to address the growing problem of drug resistant infections which is threatening modern methods to treat and prevent disease. The Welsh Government antimicrobial resistance delivery plan focuses on antibiotics to treat common infections and minor injuries, we wish to draw attention to the fact that many rare multi-system conditions require antibiotics to treat chronic bacterial infections. In our response, we explain the imperative for the antimicrobial resistance delivery plan to address the needs of the rare disease patient community.

#### The rare disease patient community

4. There are between 6,000-8,000<sup>2</sup> rare conditions which are estimated to affect 175,000 people<sup>3</sup> in Wales. Of these conditions, there are few that have effective cures or treatments. The vast majority of patients with rare conditions are left with palliation and mitigation, to limit the effect of the condition as much as possible to raise the quality and quantity of their lives. There is an enormous burden of unmet need in the community of those affected by rare conditions. Many rare disease patients must effectively combat infections, whether as a direct consequence of their condition, or due to an organ transplant or chemotherapy treatment. Without effective

---

<sup>1</sup>UK Strategy for Rare Diseases. Department of Health, published November 2013, accessed here: [www.gov.uk/government/uploads/system/uploads/attachment\\_data/file/260562/UK\\_Strategy\\_for\\_Rare\\_Diseases.pdf](http://www.gov.uk/government/uploads/system/uploads/attachment_data/file/260562/UK_Strategy_for_Rare_Diseases.pdf) (25th February 2016)

<sup>2</sup>About rare diseases, Eurordis, accessed here: [www.eurordis.org/about-rare-diseases](http://www.eurordis.org/about-rare-diseases) (25th February 2016) and The Genetic and Rare Diseases Information Center (GARD), accessed here: [www.report.nih.gov/nihfactsheets/ViewFactSheet.aspx?csid=80](http://www.report.nih.gov/nihfactsheets/ViewFactSheet.aspx?csid=80) (25th February 2016)

<sup>3</sup>Extrapolated from Council Recommendation on an action in the field of rare diseases, June 2009, accessed here: <http://eur-lex.europa.eu/LexUriServ/LexUriServ.do?uri=OJ:C:2009:151:0007:0010:EN:PDF> (25th February 2016)

antimicrobials for prevention and treatment, the success of these procedures could be compromised.

## Terminology

5. The Wellcome Trust carried out research<sup>4</sup> into public perspectives on antimicrobial resistance in 2015. One of their findings was that the term 'antimicrobial resistance' is not easy for the public to understand. Their suggested term is 'drug resistant infections', which we have adopted.

## Drug resistant infections and Cystic Fibrosis

6. An example that illustrates the impact of the current problem for rare disease patients is the use of antibiotics to treat the condition, Cystic Fibrosis (CF).
7. Cystic Fibrosis is a multi-organ disease with rapid progression which affects approximately 1 in 2,500 people from birth. Patients' lives are interrupted and restricted by the disease due to constant chronic chest infections, breathlessness, sinus and digestion problems as well as fatigue due to interrupted sleep. Prophylactic antibiotics are required to treat chronic bacterial infections, and patients may also require antibiotics if lung transplantation becomes an option due to respiratory failure.
8. The prompt use of effective antibiotics in this population has been one of the main reasons for decreased respiratory morbidity and increased longevity over several decades. Infants with CF are at increased risk of early infection and inflammation without the availability of effective antibiotics treatments. Due to the frequency of chronic bacterial infections experienced by patients with CF, drug resistant infections can be common amongst the patient population. This means that patients with CF must avoid contact with each other so ensure they do not share drug resistant infections. A necessity which poses significant problems within families and which places restrictions on approaches to treating the condition. To ensure effective treatment, clinicians may have to prescribe alternative antibiotics which can cause adverse effects.
9. The Cystic Fibrosis Trust represents patients and families affected by this chronic life-limiting condition. Keith Brownlee, Director of Impact at the Trust, said in a statement: "The significance of antibiotic resistance in people with cystic fibrosis is a matter of life and death. The effect, for people living with this condition, could be devastating."<sup>5</sup>

## Drug resistant infections within the rare disease community

10. Currently less than 5% of rare diseases have an approved treatment available. Many rare disease patient communities rely on antibiotics as a fundamental part of their treatment regime. In the example of the CF population, the quality of life, length of survival and cost of care depend on the success or failure of antibiotic treatments which have been responsible for an increase in median survival to almost 50 years.
11. There are a number of factors common to a large proportion of genetic conditions that can raise patients' risk of getting infections, or harbouring long-term chronic infections. These factors can include: compromised or absent immune systems, the suppression of an over active immune system, immune suppression to avoid graft versus host disease following transplantation, unusual skin morphology that can foster chronic infection, unusual lung morphology that can foster lung infections, and muscle weakness that makes coughing and sneezing less effective as natural defences.

---

<sup>4</sup>Exploring the consumer perspective on antimicrobial resistance, Wellcome Trust, accessed here: [www.wellcome.ac.uk/stellent/groups/corporatesite/@policy\\_communications/documents/web\\_document/wtp059551.pdf](http://www.wellcome.ac.uk/stellent/groups/corporatesite/@policy_communications/documents/web_document/wtp059551.pdf) (25th February 2016)

<sup>5</sup>Antibiotic resistance: a matter of life and death, Cystic Fibrosis Trust, accessed here: [www.cysticfibrosis.org.uk/news/antibiotic-resistance-a-matter-of-life-and-death](http://www.cysticfibrosis.org.uk/news/antibiotic-resistance-a-matter-of-life-and-death) (25th February, 2016)

12. Genetic Alliance UK supports the Welsh Government's commitment to tackle the growing problem of drug resistant infections for Welsh patients through the actions within the delivery plan. However, the plan does not take into account the use of antibiotics within the rare disease patient community and any special considerations that might be necessary. We look forward to working with the Welsh Government to address this point.



Alastair Kent OBE  
Director