



## Key Statistics from the RDUK Report 'Experiences of Rare Diseases: An Insight from Patients and Families'

### Diagnosis

- 46% of patients with a rare disease had to wait over one year for a correct diagnosis following the onset of symptoms.
- One in five (20%) had to wait over five years for a final diagnosis.
- Over one in ten (12%) had to wait over 10 years for a final diagnosis.
- Over two thirds of patients (68%) saw three or more doctors before a final diagnosis was made.
- Over one in five (22%) saw six or more doctors before a final diagnosis was made.
- 46% of patients were given at least one incorrect diagnosis before receiving their final diagnosis.
- 30% of patients received three or more misdiagnoses.

"Many doctors had no idea and some even said the symptoms were in my head" – *patient with myasthenia gravis*

"Many incorrect diagnoses - but the worse diagnosis doctors (especially GPs) give is there is nothing wrong with your child - perhaps the problem is with you (the parent)." - *Mother of a child who died from haemophagocytic lymphohistiocytosis*

### Patient Information and Support

- Over half (52%) of patients or families felt they weren't given enough information on their condition following diagnosis.
- 52% of patients or families reported that patient organisations are their main source of information on the condition.
- 37% of patients do not have someone they can go to with questions on their condition.
- Only 33% of patients feel they receive sufficient support with their social needs.
- Only 29% feel they receive sufficient support with their psychological needs.
- Only 24% feel they receive sufficient support with their financial concerns.

"Support in all areas has been poor. Benefits are now coming in because of constant pressure of me and my parents" – *Patient, Becker muscular dystrophy*

"Our son's condition is ultra rare so you HAVE to educate yourself on the topic, no doctor has time/inclination to research orphan syndromes for you." - *Parent of a child with Lowe Syndrome*

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#### Rare Disease UK

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A charity registered in England and Wales (no. 1114195)  
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“When we heard about our condition we were left to deal with it. I saw an article in paper about a lady who had the same condition as me and there was a number to ring which was Ataxia UK. I can’t thank them enough for bringing us out of the woods” – *Patient, familial hemiplegic migraine type2 with cerebellar ataxia*

“I was given a two or three sentence outline of my condition and then everything else I’ve had to research on the internet and through support groups set up by patients” – *Patient, birdshot chorioretinopathy*

“[We received a] phone call for diagnosis on New Year’s Eve with the comment from GP ‘I don’t know anything about it go on the computer and look it up’. We didn’t have a computer!!!” – *Relative of a patient with myasthenia gravis*

## Coordination of Care

- 75% of patients do not have a dedicated Care Coordinator or Care Advisor.
- A quarter (25%) of all patients have to attend either 3 or 4 different clinics for their condition.
- Over one in ten (12%) patients attend more than five different clinics for their condition.
- Two third (66%) of patients that attend clinics have to travel for over an hour to get to their furthest clinic.
- 32% of patients have to travel for over two hours to get to their furthest clinic, and 15% travel for over 3 hours to reach their furthest clinic.
- 30% of patients reported experiencing problems in the transition from paediatric to adult services. Problems were experienced in all aspects of care – medical, psychological, financial and social.

“I was ‘forgotten’ about by medics when I turned 18” - *Patient with Ehlers-Danlos Syndrome, Postural Orthostatic Tachycardia Syndrome, Raynauds Syndrome*

“There was no transition help at all” - *Relative of a patient with ichthyosis*

“Parents who care 24/7 should have more readily accessible help when needed, and respect for their care and views” - *Mother of a patient with Prader-Willi syndrome*

“If anyone would coordinate my daughter's care it would be wonderful as I've been doing it for years” - *Parent of a patient with 1q21.1 microdeletion*

## Research

- Only 35% of patients or families are informed of clinical trials into their condition.
- Only 2% of patients would not welcome the creation of a registry for clinical information for their condition.
- Only one third (33%) of patients or families feel they are provided with enough information on research into their condition.

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"We only find out because we are a member of a charity that funds research. If we were not a member of this we wouldn't know about trials etc" - *Relative of a patient with cerebellar ataxia*

"I would be very willing to participate in trials or research" - *Patient with Langerhan's cell histiocytosis*

## Access to Treatment

- Only 35% of patients were aware of a licensed treatment for their condition.
- Of those, 89% had been able to access the treatment.
- Patients and families experience inconsistencies in access to medicines, and trying to gain access to treatments can be distressing for some patients and their families.

"The delay in accessing [the drug] caused a 5 month delay in my husband's return to work. Now we are fighting for benefits as his pay has run out which wouldn't have happened without the delay" – *Wife of a patient with multiple myeloma.*

"It took over a year of negotiation by our GP and ourselves before our son's neurologist would agree to prescribe [the drug]" – *Parent of a child with Niemann Pick type C*

"I have to give my members details to take to their GP or consultant to enable him to write a script" – *Patient with systemic mastocytosis and leader of the UK Mastocytosis Support Group*

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