Dual Deprivation: Understanding the Psychological Burden that Coexists with the Physical Struggles in Individuals with Rare Diseases and their Caregivers

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Abstract

I was fortunate enough to gain an insight into what a life with a rare progressively disabling genetic condition entails. Initially fascinated with the genetics and biological mechanisms of the condition and astounded with the physical ailments the patients affected face, I soon realised this is only the tip of the iceberg. A significant number of patients with rare diseases and their caregivers face constant difficulties with their mental health, which are unfortunately often overlooked. Though this could be partly blamed on biomedical dominance, the rare nature of these conditions often mean that physical symptoms are not adequately addressed. As doctors and medical professionals, we want to fix people, after all it is our duty. When we feel this is beyond our capabilities, we almost intrinsically dismiss that patient from our thoughts, somewhat as a coping mechanism so as to not feel like we are failing. We rationalise this by telling ourselves that everything *must* have been tried already, when in reality there is always a strategy that could be implemented. It is indisputable that these patients require more from us to enable them to navigate the physical and psychological struggles they face. There is a need to raise our sensitivity to all aspects of their care to allow better detection of problems and to deliver the holistic care we would one day hope to receive.

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I was introduced to SR (pseudonym), an 18-year-old young man, and his mother NH (pseudonym) through FOP Friends[1]; a charity that supports research into Fibrodysplasia Ossificans Progressiva (FOP). FOP is an ultrarare progressively disabling genetic condition affecting 1 in 2 million people. With FOP, connective tissue such as tendons, ligaments, and muscles gradually turn into bone. The condition usually manifests in childhood and those affected often describe it as having a healthy mind locked inside a frozen body. They live their lives fearful of minor injuries or infections, which are known to trigger flare ups. They suffer from skeletal deformities, stiffness and chronic pain; all of which seriously impact their quality of life. The specific pathogenesis of FOP is not well understood and there is no effective treatment, though medications are available for symptom relief. Sadly, the average life expectancy is around 40 years[2].

SR and NH discussed several barriers to accessing services and how it fell to NH to coordinate her son's care. She spoke about her difficulties navigating the healthcare system, often getting no responses from service providers or having to travel several miles for specialists' input. Regarding consultations, they felt information was seldom shared effectively between specialists and local services. "Even if they are, sometimes they don't bother reading his file", she explained. She highlighted her relentless fights for funding, in an increasingly stretched NHS, whether this is for SR's personal care, mobility, or off-label use of drugs in attempts to alleviate his symptoms. This was only exacerbated during his transition from paediatric-centered to adult-oriented care. It was evident that there was a lack of a holistic patient-centered approach to SR's management. Though biomedical dominance is partially to blame, the rarity of FOP means that inadequacies are felt even with the management of physical symptoms.

The chances of coming across a patient with FOP are admittedly extremely low, but rare diseases as a collective are common. Nearly 8000 rare diseases have been identified, affecting an estimated 1 in 17 people[3]. Therefore encounters such as this in one's medical career are inevitable. Clinical presentations certainly differ, but the emotional burden they pose on

individuals and their caregivers is comparable. I will attempt to highlight key lessons I have learnt from SR and NH, taking into account discussions I have had with a number of medical personnel and the FOP Friends charity.

The demoralised patient

FOP creates pain and pain is demoralising. It creates disability, bodily disfigurement, social isolation, a medium for being bullied, feelings of dependency, perceptions of being a burden – all of which are also demoralising. The susceptibility to demoralisation differs between people, but even the most resilient have their breaking points. Demoralisation in patients with rare diseases seems to encompass the fundamental psychological elements of disempowerment and subjective incompetence, for example through the inability to function at one's previous or expected level. It creates a sense of futility through knowing their medical situation and by virtue the functional implications are never going to convalesce. Consequently, patients feel hopeless and helpless[4,5].

It is vital to make the distinction between demoralisation and depression. Though similarities exist including disturbances in appetite, sleep, energy levels, and even suicidal ideations, in demoralisation there is usually preserved responsivity of mood. That is as adversity ceases, the patient's capacity to feel enjoyment and to hope is quickly restored. This is not to say that demoralised patients cannot develop depression and/or anxiety. 69% of rare disease patients in the UK reported depression while 82% reported anxiety and stress[6].

Unlike depression, however, demoralisation fails to show sufficient improvement with antidepressants. Instead, demoralisation is best tackled through mitigating the patient's physical or emotional stressors as well as fortifying their resilience. With chronic conditions, demoralisation often lessens as novel management plans are delineated and symptoms are actively managed. With terminal conditions, demoralisation can be diminished when the patient believes their doctor understands their concerns, acknowledges their suffering and is capable of addressing their problems. Exploring attitudes towards hope and meaning in life, fostering search for a renewed purpose, and using cognitive behavioural therapy to reframe negative beliefs can be valuable. Some may additionally benefit from spiritual support or family meetings to enhance

family functionings[7,8]. During SR's last rheumatology review, he was told "there is nothing more I can do". The demoralised patient needs a physician who perseveres, as Cassell[9] puts it "there is *never* a time when *nothing* can be done."

Caregivers need care too

As a single parent of two, NH serves as SR's primary caregiver. During our conversation, she emphasised the daily stress and frustration she faces and the need to always "try and hold it together". This is compounded with the continuous need to advocate for her son to receive the care he deserves. Although recently matters have improved for FOP patients and their families in the UK through the development of a FOP team at the Royal National Orthopaedic Hospital (RNOH) in London, the reality is, due to financial, logistical or pain issues, help is not always sought. It becomes somewhat of a postcode lottery. Even if patients are seen by specialist services, care is not "joined up" and referrals to various services tend to be adhoc as issues arise, not through regular arrangements. A multidisciplinary team (MDT) approach can combat this issue. Regular meetings between various medical and non-medical personnel to review patients can ensure everyone gets equal access to services. Though patients differ, lessons learnt from one patient can be applied to another.

NH's duties entail coordinating her son's numerous and multifaceted needs, sacrificing hers in the process. As a parent, it is heart-rending to watch your child suffer whilst feeling like there is nothing more you can do. It is extremely distressing to think about the future; not knowing whether being a caregiver is a role which will span the course of your child's life or your own. The agony of dealing with your child's death versus the hardship your death will bring to your child.

There is guilt over how well you are meeting the needs of other family members. This unfortunately translates to using respite hours to care for them instead of yourself. Sleep deprivation, chronic fatigue, and aches and pains are just some of the physical consequences. Life is anxiety-ridden, your control of day-to-day events lost, and your ability to provide for your family diminished. Sadly, NH is not alone; 65% of rare disease caregivers in the UK reported they experienced depression while 88% reported anxiety and stress[6]. What we as doctors or

medical professionals can do to alleviate the heart-break caregivers experience is arguably very minute, but we need to exhaust all options, using a biopsychosocial approach, to make a difference.

From boy to man: transitioning from paediatric to adult-oriented care

The chronic nature of FOP, like many rare diseases, means that continuity of care is essential in minimising complications and improving outcomes. Therefore, structured transition programmes aimed at effectively moving young adults, such as SR, from paediatric to adult services are crucial for closing gaps in care. It also diminishes loss of access to allied healthcare professionals previously available to patients, and reduces loss to follow-up rates. We must address additional issues that arise from adolescence including susceptibility to mental health deterioration, independent health care behaviour, self-advocacy, self-esteem, sexual health, and advice on education and vocational planning.

Final thoughts

I was privileged enough to be exposed to what is a snippet into the lives of FOP families. Though there are a number of issues that need addressing, it is important to acknowledge the incredible work done by the FOP team at RNOH, FOP Friends, GPs, local services, etc.

Picker Institute nicely summarises patient-centered care into eight principles[10] which are seen as integral parts of providing high-quality healthcare. Those include: (1) respect for patients' values, preferences and expressed needs, (2) coordination and integration of care, (3) information, communication and education, (4) physical comfort, (5) emotional support and alleviation of fear and anxiety, (6) involvement of family and friends, (7) continuity and transition, and (8) access to care. Upon reviewing these principles, it becomes apparent why SR and NH felt their care was inadequate.

When considering the different ways rare diseases can impact affected families, it is not surprising that they encounter problems with mental health. It is almost irrefutable that additional resources are required to enable them to navigate the physical and psychological struggles they face. Acknowledging suffering, understanding concerns, helping coordinate care,

allowing for respite care for caregivers, ensuring a smooth transition to adult services are some of many strategies we can employ.

Towards the end of our conversation, I thanked NH for sharing her story with me. I remember saying: “He’s very lucky to have you”. Her response shocked me, I could hear her voice break, “You’re going to make me cry!” she replied. Of course SR is very lucky, simply acknowledging that helped build rapport between us. Sometimes all you need to do is be human!

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