EURORDIS
RARE DISEASES EUROPE

EURORDIS Guide:

Helpful Hints on Delivering a Rare Disease Diagnosis

Helpful Hints on Delivering a Rare Disease Diagnosis

Introduction

The diagnosis of a rare disease is a critical milestone for families and individuals affected by rare conditions. While it marks the end of a journey of uncertainty and anxiety, it can quickly be replaced by significant feelings of fear, stress, and guilt.

The way a diagnosis is communicated can have profound effects. When delivered with empathy and clarity, a diagnosis can empower families and individuals to face the challenges that living with a rare condition brings. However, when handled poorly, it can exacerbate emotional distress, impact our relationships and disrupt our lives. All too frequently too many in the rare disease community experience inadequate communication during diagnosis. The impact of poor communication of a diagnosis can cast a shadow across the future healthcare pathway and erode trust in all medical professionals.

A diagnosis of a rare condition is a significant and difficult milestone for the patient and family. It is also a complicated time for the clinician.

Whilst there is no good way to deliver difficult news, some ways are worse than others. This document provides a summary of some helpful hints from experts in rare diseases on how to tailor communication, information and support when delivering a diagnosis of a rare condition.

Communication:

- Take your time to deliver a diagnosis and choose in a private, quiet setting to avoid interruptions
- Deliver a diagnosis in person and always to a person affected/parent who has someone there to support them e.g.: partner, both parents and/or friend.
- State the purpose of this consultation and the timeframe available
- Use clear, non-medical language to avoid stigmatizing terms (e.g., malformation, anomalies) that may discourage open dialogue.
- Be balanced in explaining what is possible and not solely on the limitations associated with the rare condition.
- Foster a two-way conversation by providing information and checking for understanding, asking questions like, “Does it make sense?” or “Is there anything else you need?”
- Approach interactions with openness and curiosity, leaving space for questions and reactions. Ask reflective questions like, “Do you recognize any parts of the information I have given?” or “What was your reaction to hearing this?”
- Receiving a diagnosis by phone is, also for this reason, not preferable, since a person receiving the call may not be in a private setting or together with important others.
- Be mindful of your body language and observe the patient’s response, adjusting your approach as needed based on their comfort.
- After giving a diagnosis, ask, “What questions do you have?” or “how can we support you and your family?” to empower patients and offer them some control.

- Balance factual delivery with empathy to acknowledge the human aspect of the experience and the impact of the diagnosis on the family.
- Consider integrating new information and diagnoses with pre-existing conditions, diagnoses and treatments, both in the physical and the mental domain.

Information:

- Individuals affected by rare condition frequently experience a profound need for high-quality information, driven by the uncertainty associated with their condition and a need to regain control. However, the progression of rare conditions varies, making individual prognoses unpredictable.
- Remember there is no "average" person living with a rare condition—each case is unique. Provide timely, sensitive, and tailored information based on the individual's needs.
- For treatable conditions, focus on treatment options; for both treatable, chronic and progressive conditions, emphasize symptom management, clinical trials, and support networks.
- Explain the strengths often exhibited by those living with rare conditions, to give a holistic, well-rounded picture to support hope for the future life.
- Present information in a clear, understandable, and positive manner. Ensure written information is readily available and provide immediate support from staff for practical assistance.
- Use psychoeducation to explain the diagnosis and treatment; while you don't need to be a psychologist, it's helpful to understand how to communicate physical and mental details effectively and provide them with coping strategies and equipping them with skills to manage symptoms.

Support:

- Help families navigate uncertainty by sharing what you know, offering resources, and connecting them with relevant resources, patient groups and centres for expertise, be it national or international.
- Approach the diagnosis as an ongoing, possibly intense and time-consuming process with multiple interactions and offer access to psychologist immediately after the diagnostic announcement appointment and arrange a routine follow-up appointment.
- Offer reassurance, even if you don’t have all the answers immediately. Let patients know that you're working on it and will provide updates as new information becomes available.
- Try to estimate whether the process towards diagnosis or getting the diagnosis itself has been or is a traumatic event. If so, refer to mental health services addressing this trauma.
- Make clear referrals to specialists and support services as necessary.
- Clearly explain processes and expectations to help patients regain a sense of control over their situation.

THANK YOU:

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ABOUT RARE CONDITIONS AND EURORDIS-RARE DISEASES EUROPE

EURORDIS-Rare Diseases Europe is a non-profit alliance of over 1000 rare condition advocacy organisations from 74 countries that work together to improve the lives of over 300 million people living with a rare condition globally.

EURORDIS’ vision is a world where all people living with a rare condition can have longer and better lives, achieving their full potential in a society that values their wellbeing and leaves no one behind.

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