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A rare disease journey using grace and communication to create a team of experts

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The diagnosis of a rare disease is a harsh blow to a family and begins a long road of education and understanding for all involved. Navigating this process is time consuming and emotional. This presentation is to help others understand the process and hopefully have a smoother transition into the new normal they now face. As a mother of a son with a rare disease, I have cried at the official diagnosis, been grateful for a rare orphan drug that can save his life and then devastated to have it almost ripped away when our employer cut off our insurance. Working with a school to create a positive and safe day for your child can be overwhelming. Learning how to talk to and work with doctors and therapists is a key to creating a team that will effectively care for your child is incredibly important. All of these new situations can be handled through effective communication and a positive and gracious attitude. As a patient's advocate, we are their voice and there is a need for this voice to be heard by all those making life and death decisions when it comes to rare disease and the available drugs that can ultimately save them.

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Systemic amyloidosis: The paradigm of a rare disease on brink of change

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Systemic amyloidoses are a group of rare diseases caused by deposition of protein fibrils. This talk will focus on the change in this disease from an abandoned orphan disease with no treatment to major advances in the approaches to diagnosis, changing epidemiology and recent advances in treatment including much interest from the pharmaceutical industry in new drug development. Light chain (AL) amyloidosis remains the most frequently identified type but cardiac transthyretin amyloidosis is being increasingly recognized. Senile cardiac amyloidosis appears to be an epidemic awaiting diagnosis. Mass spectrometry using laser capture micro-dissection of a tiny amount of amyloid deposits from histological sections has enabled improved amyloid fibril typing. Understanding of proteo-toxicity of amyloidogenic precursors has paved the way for new therapeutic approaches. Developments in cardiac magnetic resonance imaging such as Eq-CMR and T-1 mapping have lead to accurate quantitation of the myocardial interstitial deposits for diagnosis and response assessment. 99mTc-DPD/PyP scintigraphy is transforming evaluation of cardiac amyloidosis. The availability of novel chemotherapy agents and better selection of patients for autologous stem cell transplantation have enabled delivery of therapy in AL with less toxicity and improved outcomes. An array of novel agents, including RNA inhibitors, stabilizers of amyloid precursor proteins, inhibitors of fibril formation and immunotherapeutic targeting of amyloid deposits are all now in clinical development offering great hope for specific and effective new therapies.

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