Prediction of sudden death in hypertrophic cardiomyopathy

Abnormalities of heavy chain myosin account for perhaps 50% of hypertrophic cardiomyopathy (HCM). It now seems that the site and nature of the aminoacid change influence survival. Why should this be? One possibility is that the substitutions interfere to a different degree in myofilibrillar organisation within the myocyte. It is probably significant that the substitution leading to a change in charge has the greatest deleterious effect on survival. Abnormal myofilibrillar organisation probably leads in turn to missapheal cells and abnormalities in cell to cell organisation producing disarray and an ideal substrate for arrhythmias. Family history remains a cheap way of identifying those with HCM at high risk of sudden premature death but a good case can be made for determining the exact gene abnormality. The understanding of how HCM affects myocyte structure will be further forwarded when the other genes unrelated to heavy chain myosin are discovered. These are exciting times for those interested in HCM.

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