Spotlight on Science
LESSON STARTERS

Manipulation of RNA splicing to correct disease mutations

Dr Linda Popplewell

Learning objectives:

By the end of this lesson you should be able to:

1. Describe how mutations can cause disruption of the transcript reading frame
2. Understand how antisense oligonucleotides can be used to skip out-of-frame exons and restore the transcript reading frame
3. Explain how this technology has been applied to Duchenne muscular dystrophy

Points to discuss:

1. Expansion of those hurdles i.e. mutation-specific, not curing disease, repeat administration, cost
2. Other strategies to develop a therapy for DMD – gene addition, gene editing
3. Treatment options for older boys who have severe muscle fibrosis and muscle wasting
4. This could lead to an ethical discussion of screening at birth for DMD so treatment can be started sooner and also pre-implantation screening, etc.