nature OUTLOOK

LYSOSOMAL STORAGE DISORDERS

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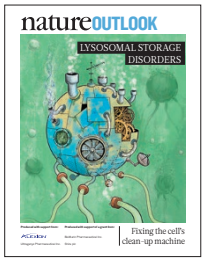

Cover art: Nik Spencer

They can come on slowly in adulthood or arrive suddenly and fatally in infancy. Their symptoms can include seizures and dementia, enlargement of the spleen and liver, and abnormal bone formation. They are rare, yet vary in frequency: the most common occur once in every 50,000 or so live births, but the rarest have been seen barely a handful of times. Indeed, at first glance, there is seemingly little to connect the 50 or so lysosomal storage disorders (LSDs), except that they all involve the lysosome (S146).

What a piece of work is a lysosome! For decades, it was thought to be just a recycling organelle — the cell's rubbish bin for disposing of unwanted cellular debris. But now it is clear that the lysosome is not only dynamic but also vital to the health of the cell (S148). Lysosomal dysfunction is implicated in other common diseases, with LSDs providing a window into the underlying cellular processes (S160).

So far, only ten LSDs have drug therapies (others are amenable to bone-marrow transplants), but LSDs with neurological dysfunction — which is most of them — are particularly poorly served. Changing this with either drugs (S154) or gene therapy (S158) is a major challenge.

Much of the support for LSD research is led by foundations set up by the parents of a child with one of the disorders (S152). Many such organizations push for the screening of newborns to identify LSDs early. Although this would seem to be a good idea, such initiatives are fraught with ethical difficulties (S162). But early detection is crucial if a person with an LSD is to have any chance of living a normal life (S151).

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Michelle Grayson
Senior supplements editor

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