



Metachromatic leucodystrophy

is a metabolic condition.

The disease is passed to the child as a recessive chromosome from two healthy parents who are both carriers of a genetic abnormality.

Abnormalities of this kind are to be found in a great many of us, but this is of no consequence, as long as they are only present in one parent.

Statistics show that if both parents are carriers of this chromosome, one in four of their children will be afflicted by the condition (there are many other varied diseases which are passed on in this way).

In the case of metachromatic leucodystrophy, the cells of the patient do not produce a specific enzyme: arylsulfatase A.

The function of this enzyme is to process certain lipids called sulfatides.

The absence of this enzyme causes an excessive accumulation of sulfatides in the cells.

These lipids progressively destroy the myelin or 'sheath' surrounding the nerves which allows for the transmission of nerve impulses.

This illness, the first symptoms of which can appear at any time up to adulthood, starts off unnoticed and then goes on to paralyse the central nervous system little by little.

In Fabrice's case the first symptoms appeared at the age of two.

His motor skills were the first to be affected, he then lost the ability to speak.

Although the progression of the disease can vary greatly from person to person, the outcome is always the same: loss of motor function, loss of the ability to speak, see, hear, ... loss of life itself

There is no currently no known cure.

If the diagnosis is made very early on (which is hardly ever possible), the only option is to attempt a bone marrow transplant.

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A.....

If you would like to become a volunteer, or partner in the Association, don't hesitate to contact us:

Tel: +33(0)5-61-67-39-91
E-mail: asso.elfe@ml.free.fr

Fabrice suffers from a rare genetic condition: metachromatique leucodystrophy.

There is currently no treatment available to help him.

But research is progressing, and all hope is not yet lost.

We want to help him to wait for a clinical cure.

For this he has to undergo a programme of physiotherapy in England.

This means four to five trips a year, each lasting a week, followed up by two to three hours of exercises a day once he is back home.

The costs generated by Fabrice's illness are already high (special equipment, the need to adapt the house ...)

Your donations will finance this programme and everything else that could improve his life on a daily basis.

THANK YOU ON BEHALF OF FABRICE

Fabrice is a little boy of 4.

He came into the world in November 2000 to the great joy of his parents.

He's a very bright child who started speaking very early, so anxious was he to ask questions about the world around him.

His parents were prompted to consult a specialist when he was slow to walk.

A condition affecting the outlying nerves was quickly diagnosed. His parents were told that this would result in a slight handicap.

But now this degenerative disease started to progress much more rapidly than anticipated, and at the beginning of 2004 Fabrice was to lose the ability to walk unaided.

Despite the unusual way in which this illness was developing, the original diagnosis was still not questioned.

By the end of 2003 Fabrice could no longer stand upright, he then began to have difficulty controlling his salivation.

At this point his parents began to have serious misgivings and started fighting to have more extensive tests carried out.

On May 18th 2004, after two days in hospital, the awful diagnosis emerged: Fabrice has metachromatic leucodystrophy.

That same week, Fabrice who had been dry for several months, needed to start wearing a nappy again.

Since then Fabrice can no longer sit unaided, he has almost completely lost the use of his hands, has great difficulty eating and can no longer speak.

Despite everything, he keeps smiling through, which wins the admiration of everyone who comes into contact with him.

www.elfe.free.fr

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Non profit-making association

ELFE

« Ensemble contre la Leucodystrophie de Fafa notre Ecrin »

« United against Metachromatic Leucodystrophy: our 'case' »

Ferme de Paraulette

09500 Mirepoix

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registered according to the law of July 1st 1901 and the decree of August 16th 1901

Tranlate by Jane

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Ensemble contre la Leucodystrophie de Fafa, notre « Ecrin »
United against Leucodystrophy for Fabrice: our case



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FABRICE

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