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# Chronic Atypical Neutrophilic Dermatosis with Lipodystrophy and Elevated Temperature (CANDLE)

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## 1. WHAT IS CANDLE

### 1.1 What is it?

Chronic Atypical Neutrophilic Dermatosis with Lipodystrophy and Elevated temperature (CANDLE) is a rare genetic disease. In the past, it has had a number of other names including: Nakajo-Nishimura syndrome or Japanese Autoinflammatory Syndrome with Lipodystrophy (JASL) or Joint contractures, muscle atrophy, microcytic anaemia, and panniculitis-induced childhood-onset lipodystrophy (JMP). Affected children suffer from recurrent episodes of fever, a rash which heals leaving bruise-like lesions, muscle wasting, progressive loss of the layer of fat under the skin (lipodystrophy), arthralgia and joint contractures. Untreated, the disease may lead to severe disability and even death.

### 1.2 How common is it?

CANDLE is a rare disease. Currently, almost 60 cases have been described in literature but there are likely to be more who have not yet been diagnosed.

### 1.3 Is it inherited?

It is inherited as an autosomal recessive disease (which means that it is not linked to gender and that neither parent needs to show symptoms of the disease). This type of transmission means that to have CANDLE,

an individual must have inherited one faulty or mutated gene from the mother and the other from the father. Hence, both parents are healthy carriers (a carrier has only one mutated copy but not the disease). Parents who have a child with CANDLE have a 25% risk that a second child will have CANDLE as well. Antenatal diagnosis is possible.

### 1.4 Why does my child have this disease? Can it be prevented?

The child has the disease because it was born with the mutated genes that cause CANDLE.

### 1.5 Is it infectious?

No, it is not.

### 1.6 What are the main symptoms?

Disease onset is in the first 2 weeks to 6 months of life. During the paediatric age, presenting manifestations include recurrent fever and attacks of erythematous, annular cutaneous plaques that can last for a few days to a few weeks and that leave residual purpuric lesions. The eye lids can appear rather thick or swollen with a reddish purple colour and the lips can look swollen.
Peripheral lipodystrophy (mainly in the face and upper limbs) usually appears towards the end of the first year of life and gets gradually worse over time.
Arthralgia without arthritis is also noted in most patients and significant joint contractures develop over time. Less common features of the disease include red eyes, inflammation of the deep tissues of ear and nose and attacks of headache due to inflammation of the brain lining. Lipodystrophy is progressive and irreversible.

### 1.7 What are the possible complications?

Infants and young children with CANDLE develop progressive enlargement of the liver and loss of both fat and muscle. Other problems, such as an enlarged heart, heart rhythm problems (arrhythmia) and joint contractures may occur later in life.

## 1.8 Is the disease the same in every child?

All affected children are likely to be seriously ill. However, the symptoms are not the same in every child. Even within the same family, not every affected child will be equally ill.

## 1.9 Is the disease in children different from the disease in adults?

The progressive course of the disease means that the clinical picture in children may be a bit different to that observed in adults. Children present mainly with recurrent episodes of fever, stunted growth, unique facial features and rash. Muscle atrophy, joint contractures and peripheral lipodystrophy usually appear later in childhood and into early adulthood. Adults may even develop dangerous alterations in heart rhythm and an enlarged and poorly functioning heart.