PATIENT JOURNEY GENERALIZED LIPODYSTROPHY

Age of diagnosis varies – usually by 10 LD has been diagnosed or misdiagnosed

1 SYMPTOMS

- Failure to thrive
- Extreme hunger
- Diabetes
- Absence of body fat
- Strong/muscular or unusual appearance in babies

2 FIRST CONTACT WITH FAMILY DOCTOR/ PEDIATRICIAN

- Suspected type 1 DM or anorexia/ eating disorder if adolescent?
- Failure to control diet blamed on poor adherence?
- Can be caught at this early stage if physicians are familiar with the disease

They think its my fault. I feel so alone

SUPPORT FROM PATIENT GROUPS

Support form patient advocacy groups is vital

3 REFERRAL

Refer to specialist, most commonly:

- endocrinologist– usually pediatric,
- diabetologist if older
- psychiatrist

This is because of your poor nutrition

Patients would benefit from help with; diet, exercise and medication

4 1ST TO 3RD LINE MEDS

- Oral anti-diabetics
- Insulin
- Lipid-lowering therapies
- Exercise and diet

Adults

Insulin doses are often not prescribed accurately

Babies

Parents are asked to switch baby formula – viewed as a nutritional issue

SUSPECTED DIAGNOSIS

Absence of SC fat, insulin resistance, hepatomegaly, cardio abnormalities. Many possible triggers.

Misdiagnosed as eating disorder. In some cases, patients can be referred to oncology for suspected cancer or referred to dermatology due to skin conditions.

DIABETES 'CONTROL'

May be attempted without Leptin replacement therapy

Insulin doses are often not prescribed accurately

After referral to a specialist a very rocky road can unfold for patient and parents

5 DIAGNOSIS / SECOND-LINE SPECIALIST

May refer on to Centre of Excellence

Average time to get a diagnosis is any time between 3-8 years depending on country of patient.

Premature mortality for some patients

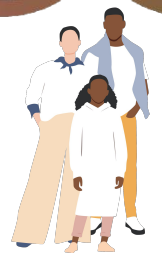
6 COMPLICATIONS

- Pancreatitis
- Liver/kidney damage
- Cardiomyopathy
- Infections
- Temperature control
- Pain as a result of no adipose tissue

7 LEPTIN REPLACEMENT THERAPY

With help from dietician Managed by CofE

I'm not hungry all the time anymore



Behavioural issues in children, cultural issues for the family as they are very embarrassed.

- Sometimes there are alternative therapies
- **Discontinuation** due to age of patient – e.g. teenagers/college - adherence
- Potentially fatal complications may ensue from years of metabolic disruption and organ damage
- Daily injections can be difficult for young patients, especially as there is complete loss of fat

8 PSYCHOLOGICAL, SOCIAL AND EDUCATIONAL SUPPORT

A clear need for psychological, social and educational support – some are lucky enough to get this

9 SUPPORT FROM PATIENT GROUPS

Support form patient advocacy groups is vital

10 THE JOURNEY CONTINUES

- Living with the long-term consequences of generalized lipodystrophy