

## **Skeletal Dysplasia: establishing metabolic & nutritional requirements to manage health risks**

### **Supervisory team:**

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### **Project description:**

Some people are born with a condition that prevents their bones from growing in the usual way. The medical term for this is skeletal dysplasia but it is often known as dwarfism. Most people notice that this results in short arms and legs but are unaware of the many other health problems that can limit quality of life (e.g. breathing difficulties due to changes in the skull, neck and back operations due to changes in the spine, joint pain where bones do not fit together well and so extra difficulties moving around in a world designed for taller people).

All the above problems can be made worse if a person becomes overweight or obese. This is important because people with skeletal dysplasia are more likely to gain weight than those who are average height. People with skeletal dysplasia are also then more likely to become ill with life-threatening diseases like type 2 diabetes and heart disease. Nobody knows why being extremely short would cause these problems. It is likely that metabolic rate might explain the links between body fat levels and health markers – but no scientific research has ever made these measurements in this unique group of people.

The reason there is no research in this area is that scientists with the necessary skills have not had access to a large group of people with skeletal dysplasia. Our research team has linked with several major charities to do just that, so we can measure metabolic rate and the metabolic response to a meal very accurately from a large number of people. We will also then use cutting-edge wearable technology to monitor diet and physical activity levels once people return to their normal lifestyles, so we can understand completely how much energy they take in and use on a daily basis.

One benefit of these measurements is that they will reveal the range of body fat that is metabolically healthy for a person with skeletal dysplasia. This is important because the usual ways to do this by simply checking the healthy range for body weight or body mass index (i.e. the right weight for your height) does not work well unless all parts of the body are the usual size and shape. Individuals with skeletal dysplasia therefore currently have no way to know whether they should lose weight and, if so, what would be an appropriate diet and physical activity for them.