

# Living with Severe, malignant osteopetrosis (SMO)

## What is Severe, malignant osteopetrosis (SMO)?

Osteopetrosis is a disease of the bones that is characterized by increased bone density. This increased bone density<sup>1</sup> results from decreased bone resorption – or the breakdown of old bone material<sup>2</sup> – due to defects in the formation and function of osteoclasts,<sup>1</sup> the cells responsible for resorption.<sup>2</sup>

**Severe malignant osteopetrosis (SMO)** is a rare congenital disorder of bone resorption that is also referred to as autosomal recessive osteopetrosis (ARO),<sup>3</sup> “marble bone disease”<sup>1</sup> or malignant infantile osteopetrosis (MIOP)<sup>2</sup> because it affects very young children.

SMO is caused by the failure of osteoclasts to resorb immature bone and characterized by frequent fractures, visual impairment and bone marrow failure. The abnormal buildup of bone tends to narrow the space inside the bone, resulting in cranial nerve, especially optic nerve, compression.<sup>3</sup> It can also result in bones that are prone to fractures and it can create less space to make bone marrow, which is where new blood cells are formed.<sup>4</sup>

## Who Does SMO Impact?

**SMO primarily affects children, with approximately one in 250,000 born worldwide with the condition,**<sup>1</sup> and from eight to 40 children born with SMO every year in the United States.<sup>2</sup> Reports of adults with SMO are rare.<sup>3</sup>

SMO is associated with a lower life expectancy, with most untreated children dying within the first 10 years of life.<sup>1</sup> Cause of death is most commonly bone marrow failure or severe infection.<sup>3</sup>

## What are the Symptoms of SMO?

Affected children usually have symptoms of SMO within the first year of life and frequently within the first three months of life.<sup>3</sup> SMO symptoms can include:<sup>2</sup>

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| ▪ Abnormal head shape  | ▪ Bone marrow failure (which can lead to anemia, bleeding problems and infections) |
| ▪ Visual impairment  | ▪ Delayed psychomotor development  |
| ▪ Hearing loss   | ▪ Delayed tooth development  |
| ▪ Rhinitis   | ▪ Inadequate tooth eruption  |
| ▪ Frequent infections (such as pneumonia, sepsis and urinary tract infections) | ▪ Frequent bone fractures  |

Talk to your healthcare provider (HCP) if your child experiences any of these symptoms.

## What Causes SMO?

SMO is an inherited disorder that results from a flaw in a gene passed down from both parents. The mutated gene for SMO is recessive and located on an autosomal (non-sex determining) chromosome, so the condition does not appear unless a person inherits the same mutation from each parent.<sup>2</sup>

A person who receives one normal gene and one gene for SMO will be a carrier for the condition, but usually will show no symptoms. A couple, both of whom are carriers of SMO, has a **25 percent chance of transmitting the condition** with each pregnancy; there will be a **50 percent chance with each pregnancy that the child will be a carrier, without SMO presenting in its lifetime.**<sup>2</sup>

## How is SMO Diagnosed?

A healthcare provider (HCP) can provide a diagnosis via an X-ray image coupled with a thorough clinical evaluation and understanding of patient history.<sup>2</sup>

- The diagnosis can be confirmed by testing the bone and by the presence of other distinct symptoms, such as vision impairment.<sup>2</sup>
- The diagnosis can also be confirmed through genetic testing.<sup>1</sup>

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## What Treatment Options are Available?

Treatment options for SMO are largely supportive and aim to help manage complications, delay progression of the disease and help patients live longer.<sup>1</sup>

- A healthcare provider may prescribe corticosteroids or high doses of calcitriol (a special form of vitamin D)<sup>1</sup> in order to slow the progression of the disease.
- Problems caused by bone marrow failure may be treated with blood transfusions.<sup>1</sup>
- Special treatments for vision, hearing and dental problems may be needed.<sup>1,3</sup>

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### References:

<sup>1</sup>Stark, Zornitza and Ravi Savarirayan. "Osteopetrosis." *Orphanet Journal of Rare Diseases*. 4.5 (2009): n. pag. Web. 29 October 2015.

<sup>2</sup>"Osteopetrosis." *National Organization of Rare Diseases (NORD)*. National Organization of Rare Diseases (NORD), 1987. Web. 29 October 2015.

<sup>3</sup>Wilson, C J and A. Vellodi. "Autosomal recessive osteopetrosis: diagnosis, management, and outcome." *Arch Dis Child*. 83.5 (2000): 449-452. Web. 29 October 2015.

<sup>4</sup>Quarello, Paola, et al. "Severe Malignant Osteopetrosis Caused by a GL Gene Mutation." *Journal of Bone and Mineral Research*. 19.7 (2004): 1194–1199. Web. 29 October 2015.