Clinical, biochemical and radiological characterization of Van Buchem disease A follow-up study

Published: 03-07-2018
Last updated: 12-04-2024

Clinical, biochemical and radiological characterization of Van Buchem Disease

Ethical review	Approved WMO
Status	Recruitment stopped
Health condition type	Musculoskeletal and connective tissue disorders congenital
Study type	Observational non invasive

Summary

ID

NL-OMON50569

Source
ToetsingOnline

Brief title
Characterization of Van Buchem disease: a follow up study

Condition

- Musculoskeletal and connective tissue disorders congenital

Synonym
hyperostosis corticalis, Van Buchem disease

Research involving
Human

Sponsors and support

Primary sponsor: Endocrinologie
Source(s) of monetary or material Support: UCB,UCB Pharma

Intervention

Keyword: hyperostosis corticalis, sclerostin, Van Buchem disease

Outcome measures

Primary outcome

Presence of VBD related clinical symptoms

Secondary outcome

* Bone turnover markers, sclerostin values in serum

* Bone mineral density (BMD)

Study description

Background summary

The rare sclerosing bone disorder Van Buchem Disease (VBD) is caused by a defective synthesis of the protein sclerostin. Sclerostin antagonises Wnt signalling in osteoblast, thereby down regulating bone formation. In VBD the decreased expression of sclerostin therefore leads to generalised hyperostosis. Due to the rarity of the disorder, literature about VBD is sparse, with only a few studies, examining small numbers of patients. A more complete characterization of this rare disorder should not only give better insight in its etiology and pathology, but can also lead to a better understanding of the working mechanism of sclerostin and its effect on bone formation.
In 2013 we reported the first cross-sectional systematic study of patients with VBD and their heterozygous disease-carriers. Our data suggested that the clinical complications of the disease stabilize with time raising the question whether sclerostin controls bone formation to the same extent throughout life. However, longitudinal data to test this hypothesis are not available either in patients with VBD or in those with the closely related disease sclerosteosis that is due to loss-of-function mutations of the SOST gene. In the present study we will test this hypothesis 8 years after the original evaluation, with specific aim to characterize their clinical, biochemical and densitometric changes with aging.

Study objective

Clinical, biochemical and radiological characterization of Van Buchem Disease

Study design

Follow up study

Study burden and risks

subjects will be asked for disease related complains, 2-3 tubes of blood will be obtained from each subject, and bone mineral density will be determined by DEXA scan.

Contacts

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Trial sites

Listed location countries

Netherlands

Eligibility criteria

Age
Adults (18-64 years)
Elderly (65 years and older)

Inclusion criteria

All patients and carriers included in the previous study aged 18 years or older

Exclusion criteria

none

Study design

Design

Study type: Observational non invasive

Masking:	Open (masking not used)
Control:	Uncontrolled
Primary purpose:	Basic science

Recruitment

NL

Recruitment status:	Recruitment stopped
Start date (anticipated):	15-06-2019
Enrollment:	40
Type:	Actual

Ethics review

Approved WMO

Date:	03-07-2018
Application type:	First submission
Review commission:	METC Leiden-Den Haag-Delft (Leiden) metc-ldd@lumc.nl

Approved WMO

Date:	18-11-2020
Application type:	Amendment
Review commission:	METC Leiden-Den Haag-Delft (Leiden)

metc-ldd@lumc.nl

Study registrations

Followed up by the following (possibly more current) registration

No registrations found.

Other (possibly less up-to-date) registrations in this register

No registrations found.

In other registers

Register	ID
CCMO	NL64854.058.18

Study results

Results posted: 15-09-2022

First publication
01-01-1900