Patient with suspected MPS II

(Early) clinical signs

Yes

Check urinary GAGs

Dermatan sulfate + heparan sulfate increased

Yes

MPS II blood enzyme testing

Iduronate-2-sulfatase low

Yes

Analyse second sulfatase

Second sulfatase deficiency

Yes

Multiple sulfatase deficiency

If molecular gene analysis available

Molecular analysis of the IDS gene

Pathological mutation identified

Yes

MPS II

No

Other MPS? Other LSD?

Iduronate-2-sulfatase low

No

Second blood enzyme test (e.g. leukocytes)

If molecular gene analysis unavailable

Second blood enzyme testing

Key

= Start/finish

= Next testing step

= Decision point