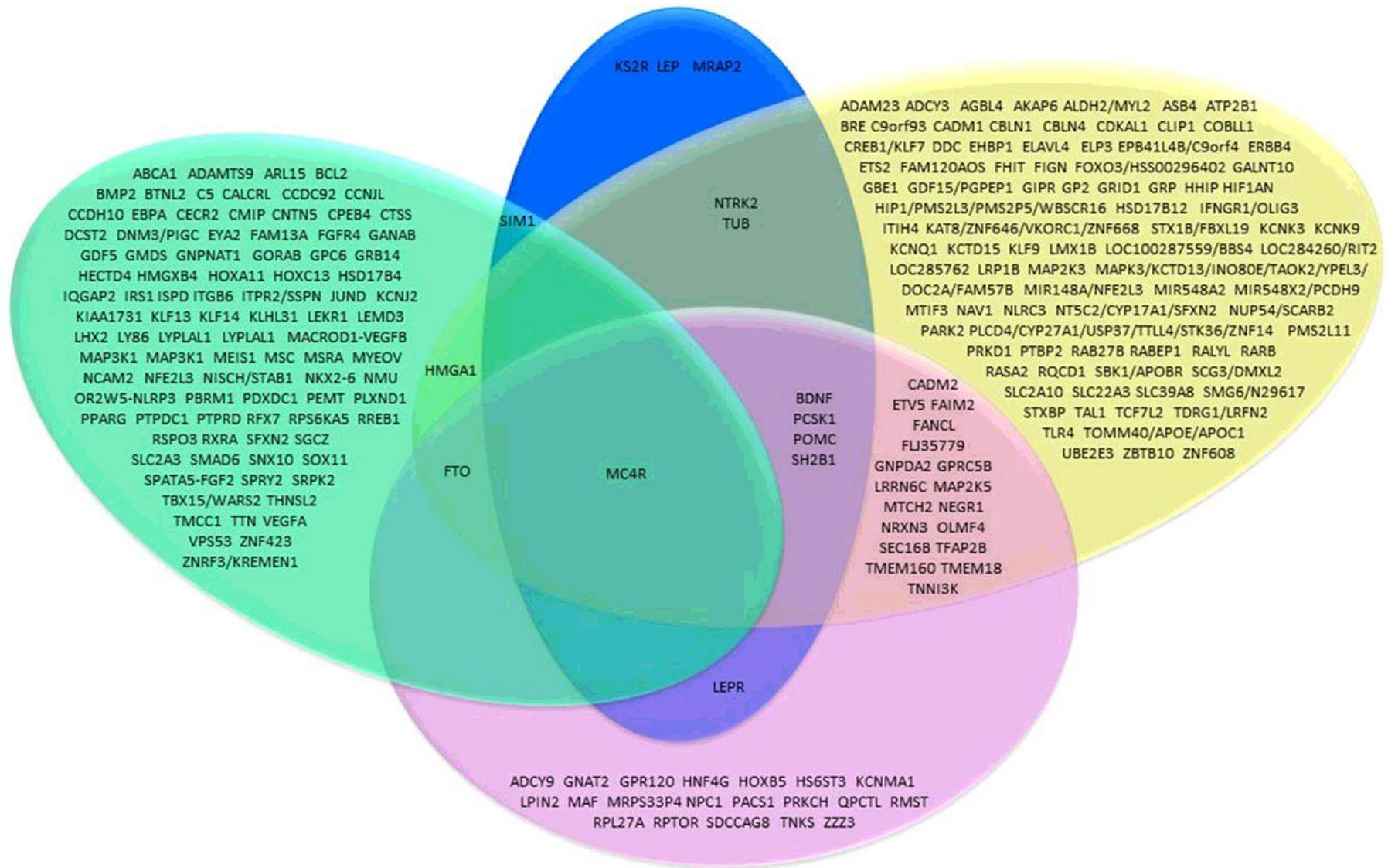


Venn diagram of genes involved in monogenic, oligogenic and polygenic obesity



Marie Pigeyre et al. Clin. Sci. 2016;130:943-986

MC4-R agonist Setmelanotide: POMC mutations and beyond..

POMC mutations:

- Severe early obesity
- Red hair
- Pale skin
- Hypocortisolism
- Delayed puberty

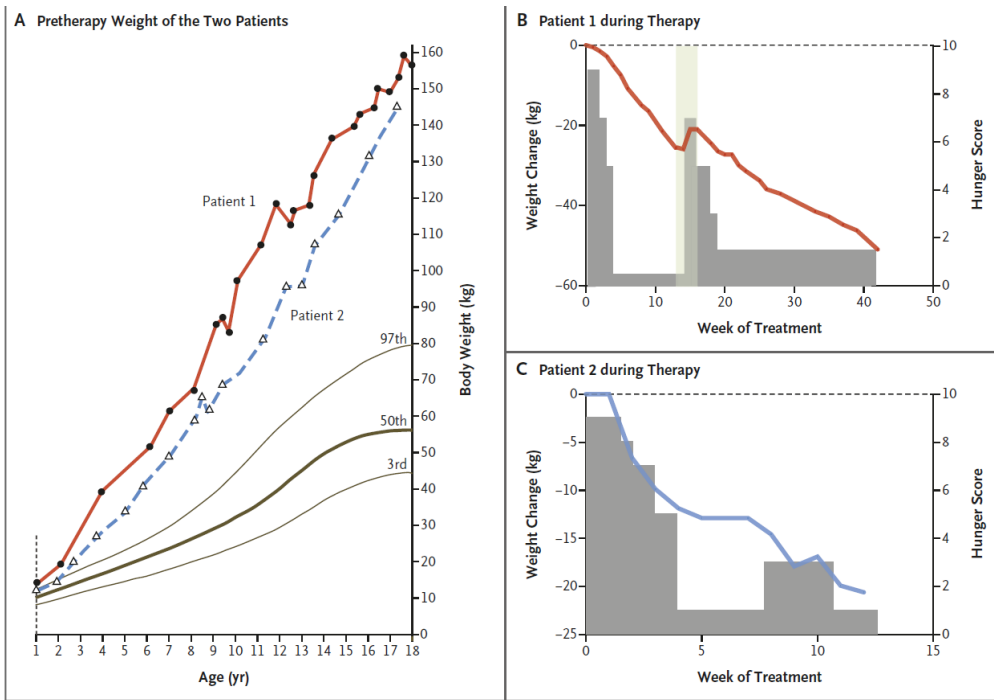


Nature Genetics 1998

BRIEF REPORT

18 years later: Proopiomelanocortin Deficiency Treated with a Melanocortin-4 Receptor Agonist

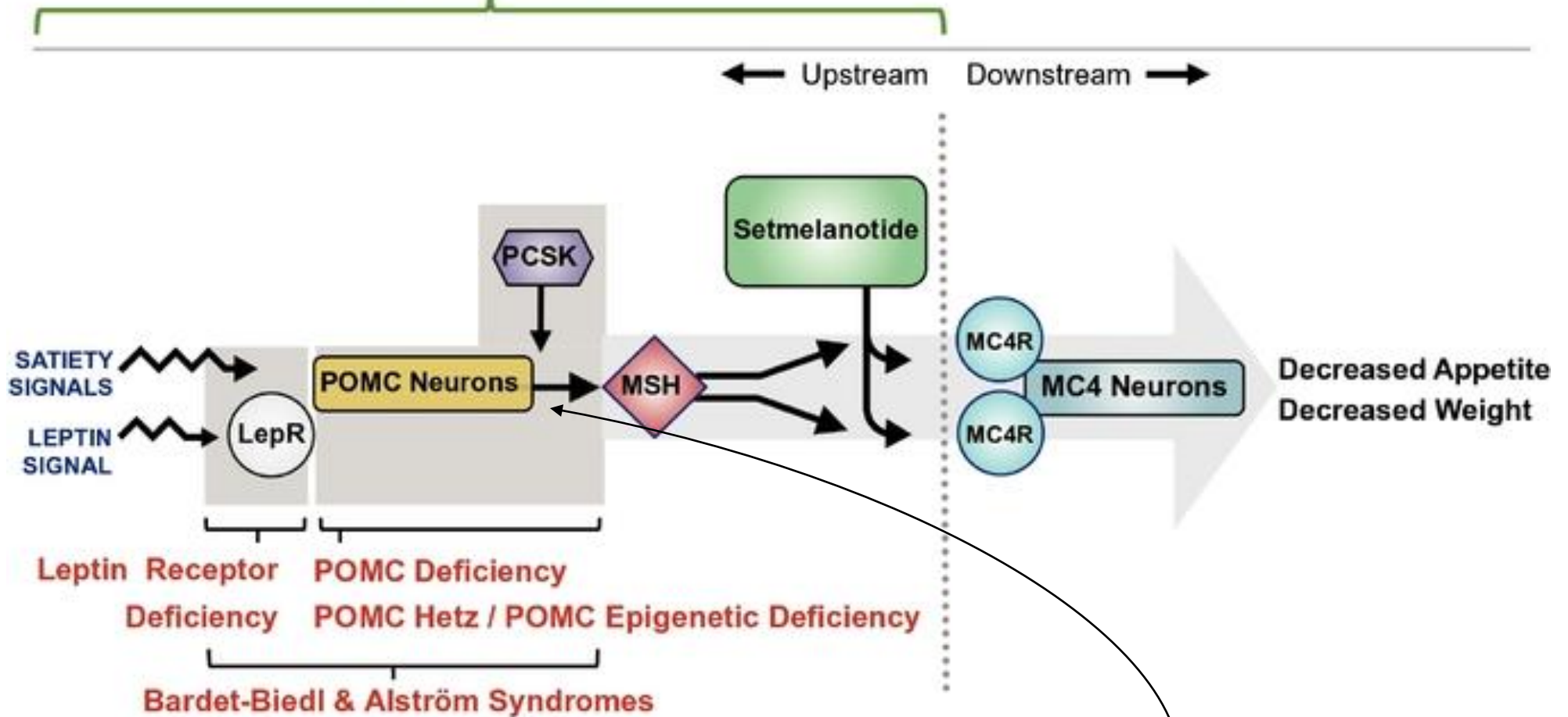
Peter Kühnen, M.D., Karine Clément, M.D., Ph.D., Susanna Wiegand, M.D., Oliver Blankenstein, M.D., Keith Gottesdiener, M.D., Lea L. Martini, M.D., Knut Mai, M.D., Ulrike Blume-Peytavi, M.D., Annette Grüters, M.D., and Heiko Krude, M.D.



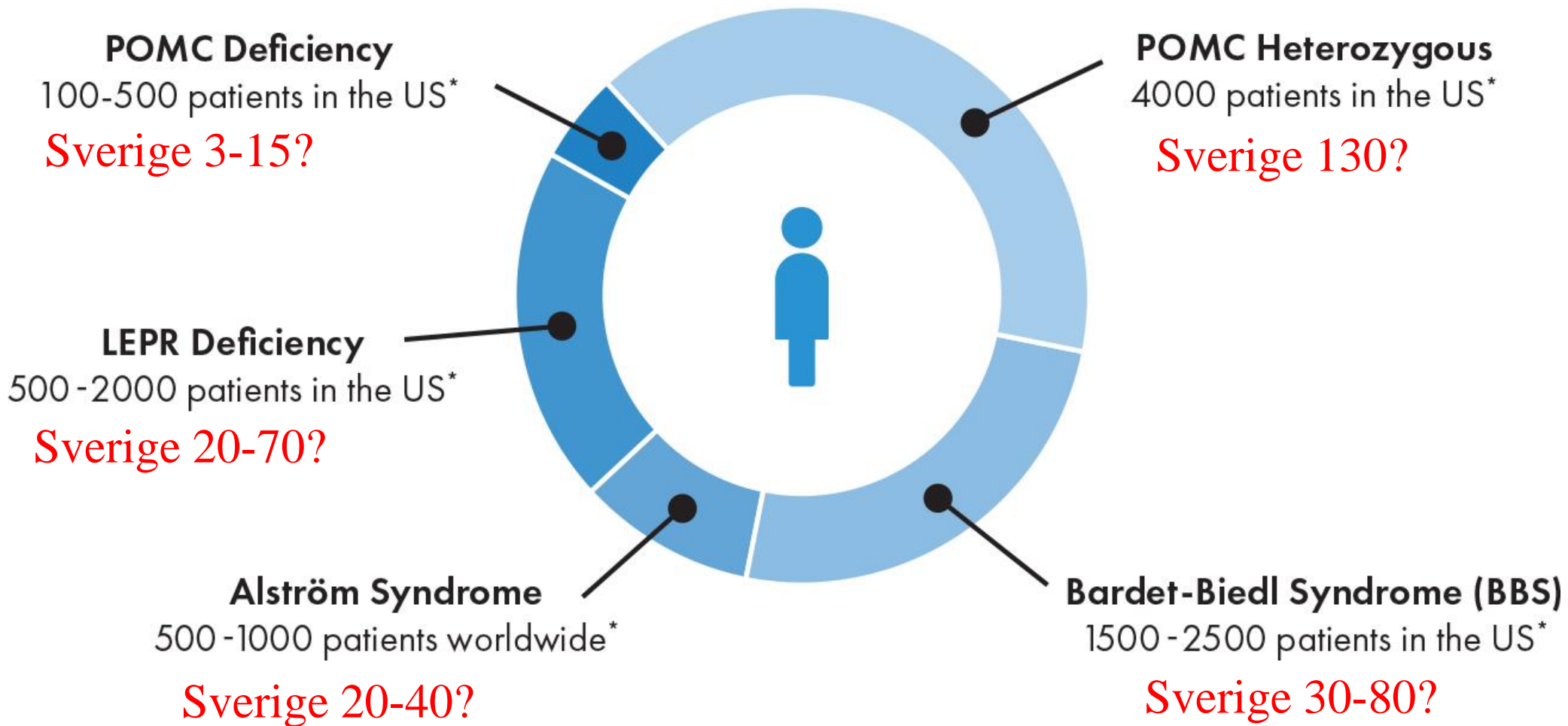
- One sc injection daily
- No remaining hyperphagia
- Increased QoL
- Darker skin and darker hair: the agonist is not totally MC4-R specific
- No negative effects on the glucocorticoid axis
- Mild adverse effect: mouth dryness, "mild episodes of fatigue, sadness or emptiness"

MC4 Pathway

Rhythm Focus



GLP-1 receptor agonister



Other: PWS (orphan drug status for setmelanotide)
Post surgery for hypothalamic tumors
Severe simple obesity

Rhythm vill screena barn med extrem fetma för mutationer i hypothalamusregionen

- Specifika kriterier: extrem fetma/tidigt utvecklad fetma/extrem hunger
- Blodprover samlas in och analyseras i USA
- Etikgodkännande krävs

Fråga: vill vi vara med?

+

Vi kan få genetikundersökningarna "gratis"

Vi kan erbjuda barn med stort lidande en möjlig behandling

-

Skicka genetiska prover till företag i USA

Minskad kontroll över analyserna

?