

IN FOCUS PRO SPECTRUM



WHAT IS PROS?

PIK3CA-Related Overgrowth Spectrum is a group of genetic conditions which cause tumour-like overgrowth of body tissues. Mutations are spontaneous and growths occur in many parts of the body including limbs and internal organs.

Conditions under the PROS umbrella include fibroadipose hyperplasia and facial infiltrating lipomatosis. Treatment includes surgery to remove overgrowths and medications to manage seizures if growths impact the brain.

MECHANISTIC SIMILARITIES

Growths in PROS are not cancerous, but PIK3CA is implicated in some breast cancers (BC). Researchers turned to Alpelisib, a drug being developed for BC. The fact that conditions are caused by the same gene suggests the same treatment could be used in both.

Researchers in France, lead by Dr Canaud, gathered mouse model data to support their hypothesis while they waited for Alpelisib to complete Phase III BC trials. Data was promising and, by working closely with the marketing authorisation (MA) holder and regulatory agencies, the team were able to conduct research into PROS.

Real world evidence (RWE) was retrospectively gathered from individuals who had been prescribed Alpelisib on a compassionate basis. Data was collated to form the evidence presented in the EPIK-P1 trial. In 2022 Alpelisib was granted an MA for use in PROS in the USA.

FORMING RELATIONSHIPS

WORKING WITH INDUSTRY

Because the MA holder owned the rights to produce and market Alpelisib, their agreement was essential. It's important to understand the motivations of industry to secure their cooperation. Building trust and matching your proposals to their goals where possible is key.

WORKING WITH REGULATORS

Seeking scientific advice before submitting evidence is a great way to ensure you are collecting the right data, at the right time, and in the right way. For Alpelisib, good communications with the FDA resulted in approval based on real-world evidence and the successful trial. This fast-tracked getting the drug to PROS patients.

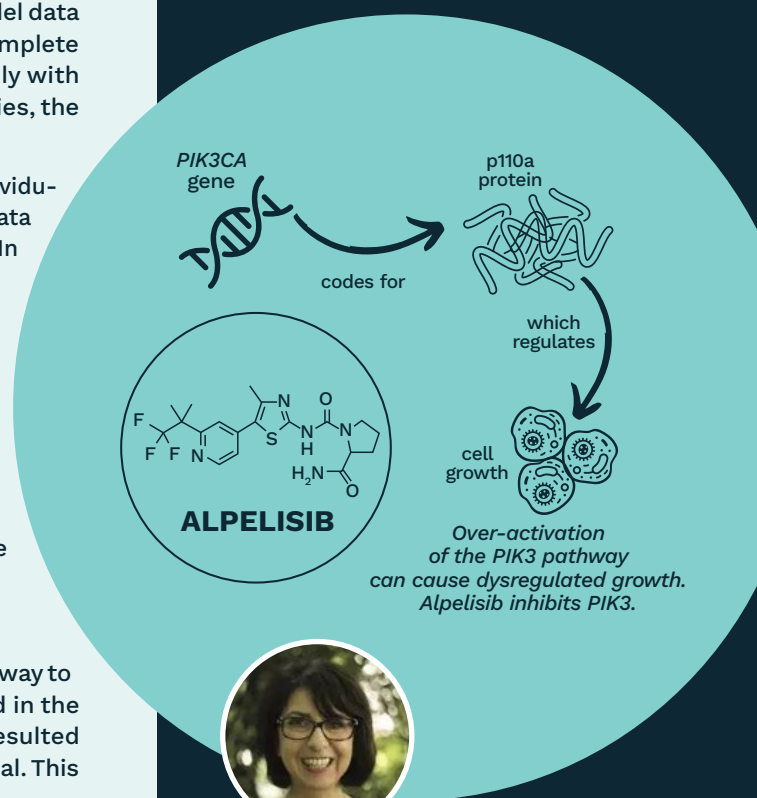
CHOOSING THE RIGHT TIME

Researchers had to wait for the MA holder to finish their Phase III clinical trial before they would consider repurposing. You may have to delay plans to apply for specific funding or wait for a company's new goals to come into effect.

AT A GLANCE

- PROS is a group of spontaneous genetic conditions.
- It causes tumour-like overgrowth of various tissues.
- The same gene that causes PROS is linked to breast cancer.
- Breast cancer drug, Alpelisib, has been successfully repurposed for PROS. Communicating with the FDA and the MA holder resulted in swift approval based on real-world evidence.

HOW IT WORKS



SPECIAL THANKS TO CHRISTINE FETRO

Christine is head of research and industrial partnerships at the Foundations for Rare Diseases in France. Her expertise in medicinal regulations was invaluable in creating this case study.



REMED4ALL is funded by the European Union's Horizon Europe research and innovation programme under grant agreement No 101057442.

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