Research in Rare Diseases - When Less is More

In this Thematic Issue, authors try to shed a light about the complexities to develop new knowledge in the field of rare tumours. All the examples share some difficulties but also have their own particularities regarding their special area of interest.

The main commonalities are related to the difficulties of obtaining sustainable funding for research in all the aspects from clinical to translational. Charities around specific diseases and patient advocates play a key role to gain awareness in these scenarios.

In the first review, Dr. Lamarca et al. focused on Biliary Tract Cancers [1]. Biliary Tract Cancers (BTCs) have been neglected by the wider cancer community, largely due to their relative rarity and the challenges of obtaining sufficient patient numbers for adequately-powered studies and sustainable funding. National and international collaboration is attempting to overcome these hurdles. BTCs are not a unique disease, but a mix of different malignancies which differ not only in molecular aspects, but also in clinical and demographic characteristics. Tailored management, smart clinical trial design, improved cross-sectional and functional imaging, and enhanced quality of life assessments are required.

In their review, Dr. Heredia-Soto et al. explained how the development of high throughput techniques has moved the biomarkers research field to a new scenario with large numbers of samples. Access to well annotated patient derived models and high quality associated clinical data is paramount for effective translational research. The establishment of new in vitro techniques alternative to the existing ones, such as, the refinement of 3D culture allows for better modelling of the architectural conformation of the tumour [2]. This is translated into more opportunities for research in rare tumours.

Dr. Frizziero et al. made a thorough review about the aspects of research in Hepatocellular Carcinoma (HCC) from clinical research to translational research [3]. In HCC, it is difficult to establish universally recognisable standards of care and clinical decision making is hampered by a number of areas of controversy and uncertainty in regard to best practice. The clinical and biological heterogeneity of HCC mandates tailoring of treatment to individual patients, at all disease stages, to achieve best outcomes. A stepwise construction of a personalised treatment plan using both loco-regional and systemic therapies in varying sequence or combination along the disease course of a single patient has become widespread in clinical practice.

Dr. Hernando et al. reviewed the state of research in thyroid cancers concluding that from translational research, an improvement in the knowledge of the less common subtypes of the DTC and more aggressive variants like MTC and ATC is urgently needed [4]. The establishment of new molecular subtypes will allow to move forward in the field of targeted therapies as well as to move from general multi-kinase inhibitors with multiple off-target toxicities to developing new highly selective drugs. In the clinical field, we should optimize the inhibition of BRAF, exploring combinations of drugs to overcome the resistance and adapting the data generated in other tumors such as melanoma or colorectal cancer, but at the same time considering the different conditions of the TCs including different sensibilities and resistances across tumors types.

The thematic issue also reviews the challenges of neuroendocrine tumour research from clinical trial design to optimization of samples [5].

All these entities share two main problems, heterogeneity within low frequency (but, sometimes relatively higher prevalence than expected) and limited resources. The scope of this thematic issue is raising awareness and help the reader to understand what sort of hurdles a new and enthusiastic researcher needs to bear in mind when initiating a research plan or a career in the field.

We expect that this thematic issue can be used as a tool to guide researchers through the most common problems when approaching the study of rare cancers. The examples showed in this issue are common to other rare diseases and rare cancers.
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REFERENCES


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