



Reflections on using Laboratory Genetic Counsellors in the Genomic Era

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Reference Laboratory Genetics

Abstract

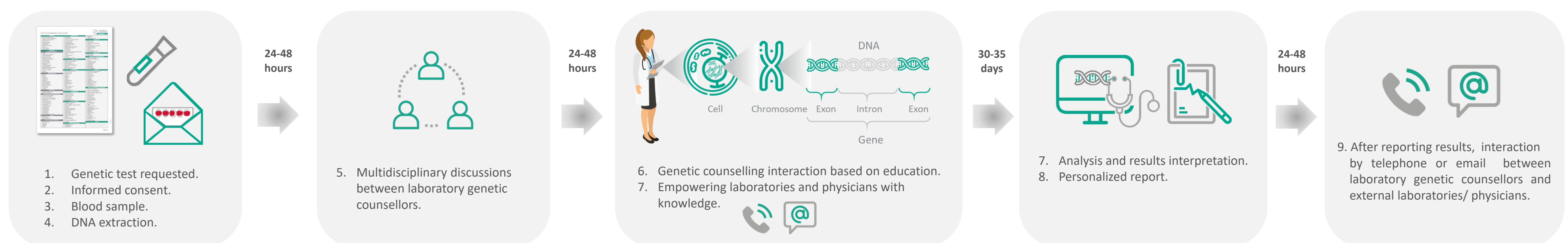
Knowing that we find ourselves in the Genomic Era, where researchers and clinicians have started to use the knowledge of genomics to improve health; the presence of genetic counsellors are becoming increasingly necessary within laboratories themselves. The current demand in both public and private centres for exomic and genomic tests shows the importance of conducting genetic counselling before and after the genetic analysis itself. Laboratory genetic counsellors are well trained in understanding different “languages” (medical terms and lab tests) in order to be an adequate and valid interlocutor between the laboratory and the clinician.

In the Genomic Era, which variants will provide and affect the results, combined with how we are going to report on them, must be at the core of respecting the autonomy of the patient. They must be based on guidelines and a specialized informed consent document. The role of a laboratory genetic counsellor in a patient-focused system avoids, among other things, the application of unnecessary genetic studies, as well as facilitation of a personalised report corresponding to choices made by the patient.



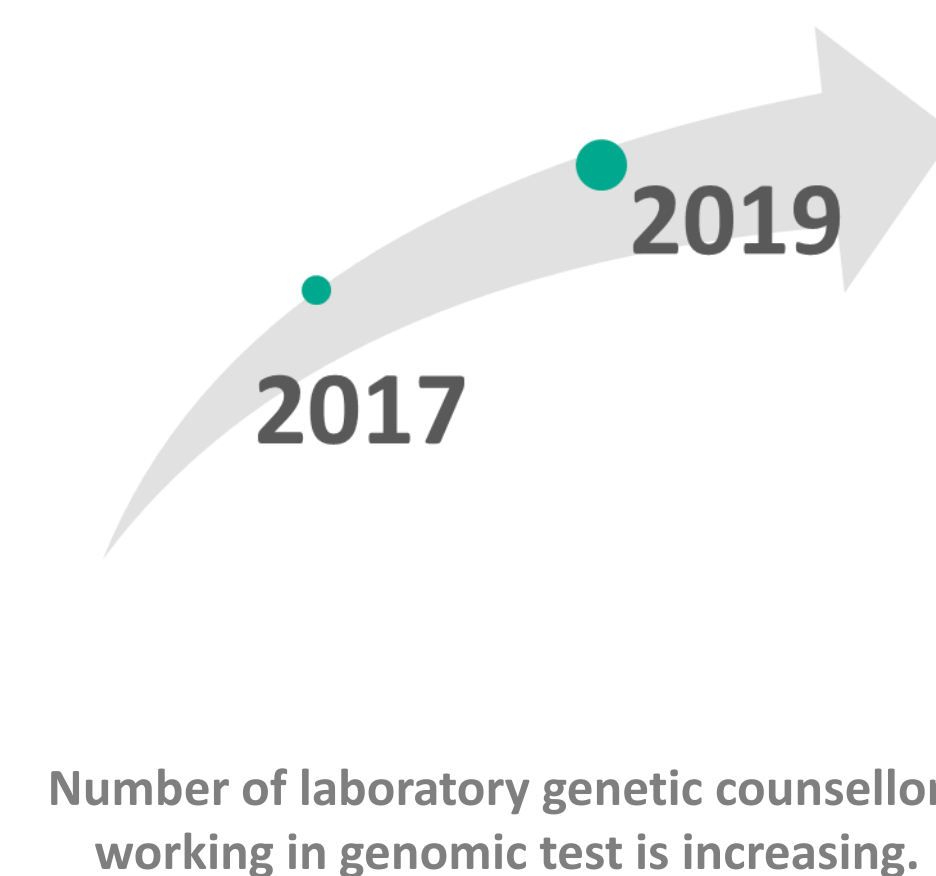
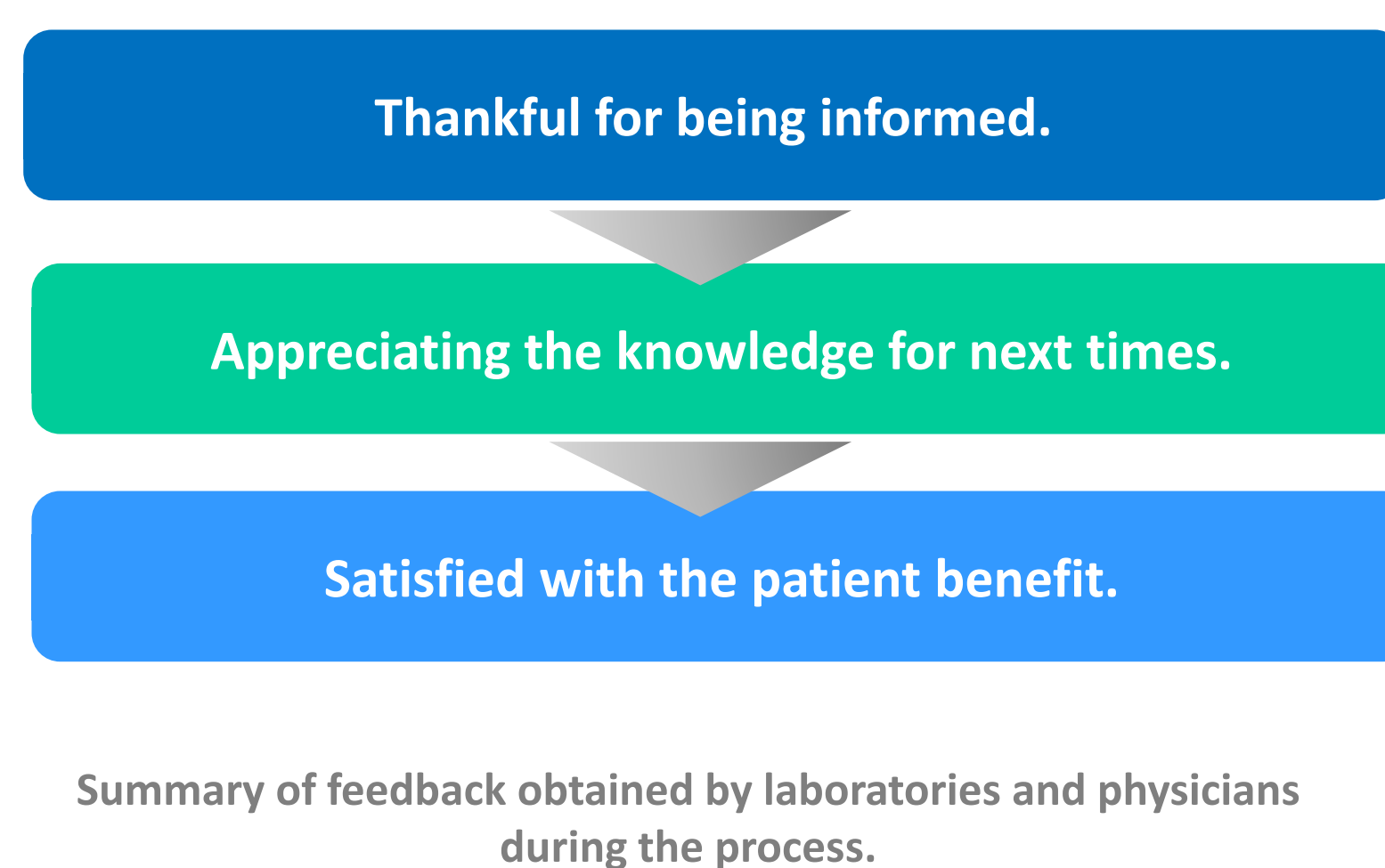
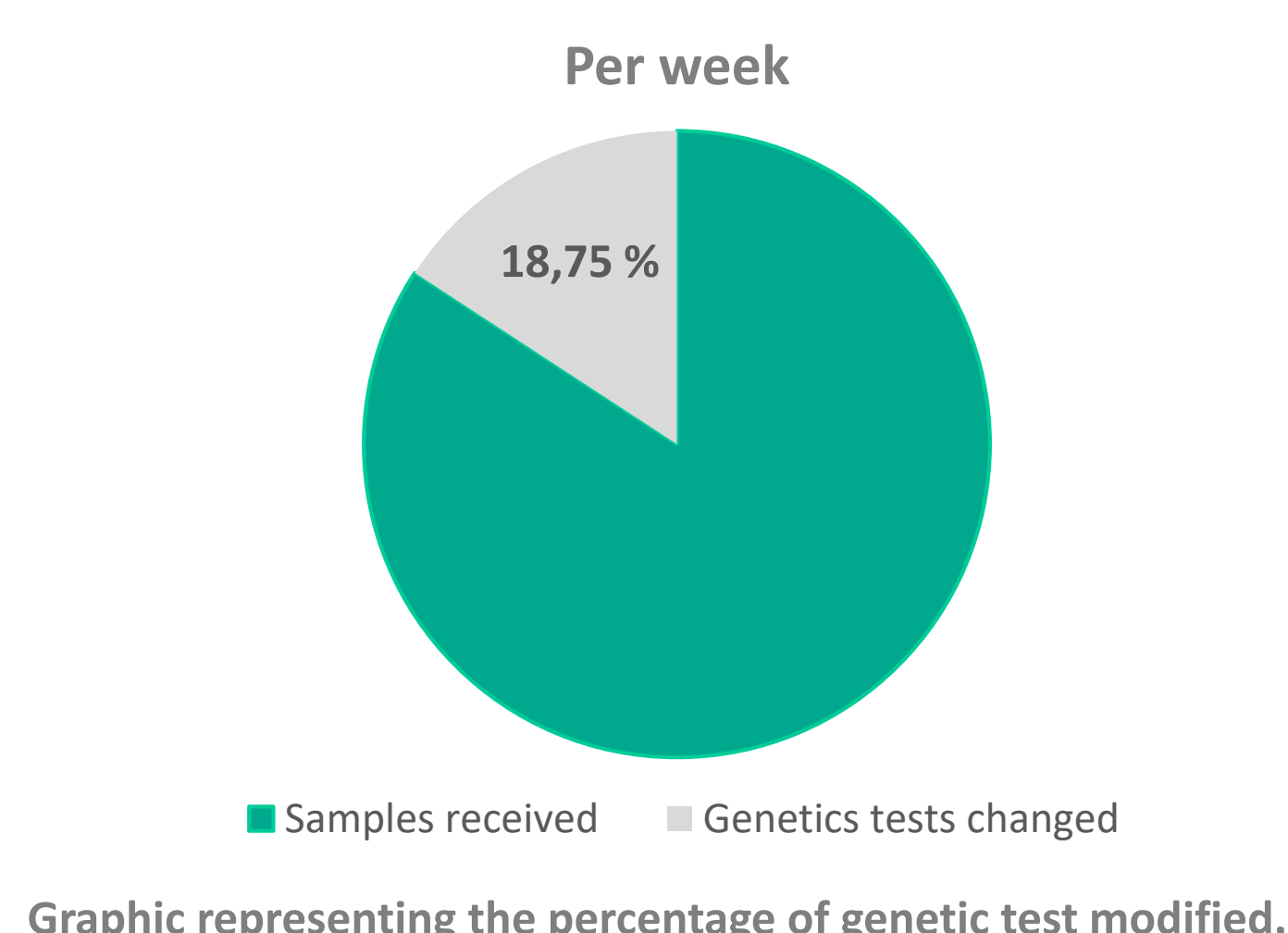
Methodology

Our centre is a genetic/genomic test accredited laboratory, putting into practice laboratory-driven genetic counselling. For that, we model our workflow experience with the challenge of the plane of communication and interaction between the clinician and the laboratory. After receiving a request form, we pre-analyse the clinical data and the genetic test that was ordered. The same process is followed before results get sent out. In case of inconsistency, the inherent skills of the counsellor are applied even when contacting the clinicians by phone or email.



Active and dynamic intervention between Reference Laboratory Genetics and other external laboratories/physicians by telephone or email

Results



A significant number of genetic tests are being changed into useful analyses. Based on feedback provided by clinicians, contacting them directly, gives them the confidence to communicate the information/results with the patient. This model of workflow applies counselling skills, particularly in the genomic era and also assists with improving the diagnostic process for families with a suspicious genetic conditions. Over, in the past two years, our team have had to double the number of staff who work in genomic testing.

Conclusions and challenges for the role of the Genetic Counsellor in the next years

PRE	POST
Anamnesis <ul style="list-style-type: none"> Detailed clinical information (HPOs). 	Genetic counselling <ul style="list-style-type: none"> Clinical geneticists. Personalised.
Genetic counselling <ul style="list-style-type: none"> We advise on the most indicated diagnostic test for the patient. Direct communication. 	Conclusive medical reports <ul style="list-style-type: none"> Validated by our geneticists. Detailed description of methods used. Differential diagnosis and detailed analysis of the clinical information received. Recommendations.
Sample processing <ul style="list-style-type: none"> Certified Laboratory. Total sample traceability (RefLabW). 	Extension analysis Variant re-classification service

Patient centred workflow

