Written Submission Template

The consultation draft of the National Health Genomics Policy Framework (the Framework) is written as an outline and poses questions throughout as the basis for consultation. These questions are focussed on the key domains for priority action.

Feedback from the consultation will enable drafting of the National Health Genomics Policy Framework to be finalised and submitted for Australian Health Ministers' Advisory Council endorsement through the Hospitals Principal Committee.

Confidentiality of submissions
Unless otherwise indicated, all submissions may be published on the Department of Health website. If you wish any information to be treated as confidential, please explicitly and clearly identify that information and outline the reasons why you consider it confidential. General disclaimers in covering emails will not be interpreted as a specific request or taken as sufficient reason to submissions to be treated conditionally. Submissions including personal information identifying specific individuals will be de-identified prior to publication.

How to submit your written submission
Written submissions on the consultation draft of the National Genomics Policy Framework can be sent in Word format by email to genomics@health.gov.au by close of business on **8 March 2017**.

1. **Full name:** Nicole Millis

2. **Email:** nicole.millis@rarevoices.com.au

3. **Are you providing your response on behalf of an organisation?**

   [Yes] If Yes, please specify: Rare Voices Australia

4. **If applicable, please specify you and/or your organisation's area of expertise**. I am Executive Officer of Rare Voices Australia, a national not-for-profit organisation that works with all stakeholders to be the unified voice and advocate for Australians living with rare disease.

5. **Do you consent to potentially being contacted to discuss the content of your submission further?**

   [Yes] Phone Number (Optional): 0459 021 204

6. **Do you accept the terms specified above about the confidentiality of submissions?**

   [Yes] If no, refer to the instructions in the disclaimer above.

Section of Framework		Question	Response
Glossary	Pg. 2-5	7. Are there other key terms referenced in the Framework which should be added to the glossary? If so, please provide details.	*Consumer, patient, stakeholder etc. Acronyms of genetic and rare disease organisations (like RVA, GARDN, SWAN, GAA, GSNV. The glossary seems to reflect a technology-centred focus rather than a patient-centred focus which is concerning for the framework's overall aims.*
		8. Are the definitions easy to understand? Do any definitions require amendment? If yes, please provide details.	*No the definitions are very scientific and not overly appropriate or aimed at the perspective of the lay person, importantly including the patient/consumer and policy maker.*
Preamble	Pg. 7-8	9. Does the Preamble provide a sufficient overview of the Framework? If not, please provide further details.	*The Preamble would be considerably strengthened if it clearly positioned the drivers of the framework as person-centred (patient/consumer centred) rather than driven purely by technology. The Preamble and Principles etc seem to be driven by the potential of the technology, instead of the needs of patients. It is unclear how the patient/consumer voice was and will be prioritised within the Framework. RVA would also like to see a prioritisation of arguably the greatest unmet need and genomic technology's potential to significantly address this ie within the undiagnosed/rare disease space.*

Section of Framework		Question	Response
		10. Are there linkages with other key frameworks or strategies that should be explicitly referred to in the Preamble? If yes, please provide details.	*International genomic frameworks should be referenced and leveraged from to ensure Australia is working towards best practice. RVA and many in the rare disease community are advocating for a National Plan/ Framework for Rare Disease. To best respond to the needs of the rare disease community, a National Genomics Framework must be aligned with and support a much needed and broader rare disease policy framework that includes not just improved diagnostics, but better access to services and clinical care, development of and timely access to treatments, improved research and data collection.*
		11. Is a three year timeframe sufficient for the Framework? Please explain your answer.	*While it is likely that 3 years will not be sufficient to undertake all that is needed, if the strategy is adequately planned with effective consultation, the work targeted at critical areas, then it could make much needed significant advances.*
Strategic Context	Pg. 10-12	12. Are the most critical international and national activities referenced? If no, please provide details of what may be included and why it is important.	*International genomic frameworks should be referenced and leveraged from to ensure Australia is working towards best practice. RVA and many in the rare disease community are advocating for a*

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			National Plan/ Framework for Rare Disease. To best respond to the needs of the rare disease community, a National Genomics Framework must be aligned with and support a much needed and broader rare disease policy framework that includes not just improved diagnostics, but better access to services and clinical care, development of and timely access to treatments, improved research and data collection.
		13. Does the Strategic Context provide a clear case for improved national consistency in genomics policy? Please explain your answer.	*The Strategic Context does provide a clear case for improved national consistency in genomics policy. To further strengthen it RVA would like the framework not just to call for consistency/ uniformity but address the issue of competitive duplication which is arguably a by product of our largely state-funded health system, but not overly responsive to the needs of the rare disease community where there is much need but only limited resources.*
An Australian Perspective	Pg. 13-14	14. Are there additional barriers, issues or challenges to integrating genomics into the health system that should be included in this section? If yes, please provide details.	*RVA suggests that these 'issues' are reworded more positively to better highlight the opportunities whilst providing context to the challenges/ barriers and potential ways forward/ to overcome these. Once again, language and principles of collaboration and patient/consumer engagement would better shape*

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			this section. *Equity of access is also a current issue, not just due to geographical reasons but cost, and CALD considerations etc*
A National Health Genomics Policy Framework for the next 3 years	Pg. 15	15. Are the key guiding principles appropriate? Please explain your answer.	*The key guiding principles are certainly not inappropriate but would greatly benefit from stronger, clearer, patient-centred language that better acknowledges the current state of play (strengths and gaps) of the health system.*
Enablers	Pg. 16-17	16. Are there additional enablers that should be included? If yes, please provide details.	*A much-needed National Rare Disease Framework*
Strategic Intent	Pg. 16-17	17. Is the Strategic Intent of the Framework appropriate? If no, what would you suggest?	*Once again, the words/ language should be re-ordered to position the framework is firstly patient/ consumer -centred rather than technology centred. This would be a simple yet important change.*
Priorities Areas	Pg. 17	18. Are the priority areas appropriate? Please explain why or why not.	*RVA would like the priority areas to acknowledge and respond to the current inequities, unmet clinical need (gaps) in the health system for genetic (rare) diseases. The potential for genomic technology to*

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			reduce the diagnostic odyssey commonly experienced by those with rare disease. The fact that early diagnosis in the gatekeeper to the best clinical care and optimum patient care, particularly when much of rare disease is life-threatening and progressive.
Overarching priority – ethical, social and legal (regulatory) issues	Pg. 18-24	19. Is the placement of ethical, social and legal (regulatory) issues as an overarching priority appropriate?	*It may be even more appropriate for this to be guiding principles for whole framework rather than a separate priority.*
		20. Should these issues be considered prior to the six priority areas, or after?	*All should be considered together.*
		21. Are there any other broad ethical, legal or social issues that should be addressed under this priority? If yes, please provide details.	*Yes engagement, facilitation and prioritisation of the patient/consumer voice, in all stages of implementation and all throughout the Framework.*
Priority Area 1 – Strong leadership and	Pg. 25-26	22. With regard to *Priority Area 1 – Strong leadership and governance*, is anything missing	*RVA thinks this is one of the strongest parts of the Framework as it currently reads. Again it would be strengthened by aligning itself with, or as part of a*

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governance		or what should change, for: (a) the current situation; (b) why is this important; (c) opportunities for improvement; and/or (d) what the future looks like?	*much-needed broader national rare disease strategic framework. Early Diagnosis is a key critical need in Rare Disease but patients also need a national response to systemic service planning, coordinated care, treatment development and timely access to this, national approach to data collection and research.*
Priority Area 2 – A skilled and literate genomics workforce	Pg. 27-29	23. With regard to *Priority Area 2 – A skilled and literate genomics workforce*, is anything missing or what should change, for: (a) the current situation; (b) why is this important; (c) opportunities for improvement; and/or (d) what the future looks like?	*The role of the consumer is missing here, in regard to shared clinician/patient decision-making, and in responding to priorities of the patient/consumer, both on an individual level and broader stakeholder level.* *Access to services is currently very inequitable. This needs to be addressed.*
Priority Area 3 – Application of genomic knowledge is evidence based, high quality	Pg. 30-31	24. With regard to *Priority Area 3 – Application of genomic knowledge is evidence based, high quality and safe*, is anything missing or what	*This is definitely a crucial priority area. Lack of data is an inherent feature of rare disease which negatively impacts on the whole rare disease xperience from diagnosis, clinical care, service*

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and safe		should change, for: (a) the current situation; (b) why is this important; (c) opportunities for improvement; and/or (d) what the future looks like?	*planning, development of and access to treatments, and research*
Priority Area 4 – Integration of genomic knowledge into person-centred health care, supported by equity of access to services	Pg. 32-33	25. With regard to *Priority Area 4 – Integration of genomic knowledge into person-centred health care, supported by equity of access to services*, is anything missing or what should change, for: (a) the current situation; (b) why is this important; (c) opportunities for improvement; and/or (d) what the future looks like?	*All previous responses focus on these areas which should be more central to the Framework itself.*
Priority Area 5 – Sustainable investment in health	Pg. 34-35	26. With regard to *Priority Area 5 – Sustainable investment in health genomics*, is anything missing or what should change, for:	*The argument for sustainability can be strengthened if clearly seen as a response to high level of unmet need (rare disease/ undiagnosed diseases). In one recent survey, 30% of Australian*

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genomics		(a) the current situation; (b) why is this important; (c) opportunities for improvement; and/or (d) what the future looks like?	*patients waited 5 or more years for correct diagnosis. 50% of Australian had one or more misdiagnosis. Diagnosis is the gatekeeper to most effective (and thus efficient) care. Reducing the diagnostic odyssey of rare disease, also markedly reduces the cost of disease, clinical cost, health cost, psychological cost, but also importantly the economic cost.*
Priority Area 6 – Effective and appropriate collection, management and utilisation of genomic data	Pg. 36-38	27. With regard to *Priority Area 6 – Effective and appropriate collection, management and utilisation of genomic data,* is anything missing or what should change, for: (a) the current situation; (b) why is this important; (c) opportunities for improvement; and/or (d) what the future looks like?	*As previously stated a national data collection system is vital, from the perspective of rare disease which is negatively impacted by a lack of data collection. In rare disease much more evidence/knowledge is needed around diagnosis and prognosis and also treatment/ best care. It is important that a system of data collection is longitudinal, contributes to broader knowledge, is translational to better patient outcomes and reflects what is important to patients.*
Implementing the Framework	Pg. 39	28. Is the suggested approach to implementing the Framework reasonable and appropriate? Please explain your answer.	*This is perhaps the sparsest part of the Framework. While this is understandable to a degree, it is vital that it sets out a plan for stakeholder consultation, particularly with the rare disease community. A*

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			better understanding of the challenges faced by the rare disease community both at the individual micro level but also at a broader policy level is important.
Overarching questions (Relates to the entire Framework)	Pg. 39	29. Is the structure of the Framework appropriate and easy to follow? Please explain your answer.	*As already outlined, the Framework seems overly driven by the technology rather than patient/consumer needs. There is a marked absence of patient/consumer voice within this framework and this should be urgently remedied.*
		30. How could the review and evaluation of the Framework be strengthened?	*I urge you to reconsider the needs of the patients/ consumers, particularly those living with rare disease, to best contextualise this framework. The Framework needs to be patient-centred and driven rather than technology – centred and driven.*
		31. Do you have any other feedback on the Framework?	*RVA is very pleased to see a national approach and would be pleased to help ensure a stronger patient voice within this Framework. RVA would also be pleased to support the implementation of this Framework, as part of its work towards a broader National Framework for Rare Disease.*
		32. Are there any issues you would like covered at	*Stronger consumer voice, engagement and*

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		the stakeholder consultation forums in February 2017?	*prioritising of the consumer experience – particularly that of rare disease.*
Genomics Framework One Page Outline and the Companion Document		33. Do you have any feedback on the Genomics Framework One Page Outline (noting that it provides a summary of the Framework) or the Companion Document?	*Just the point, I often find myself making – any focus on the benefits of personalised medicine should always be tempered by acknowledgement of the reality of rare disease, that for the majority of rare diseases, there is no medicine let alone 'personalised' medicine.*