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:** Heather Renton

2. **Email:** president@swanaus.com.au

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

   [Yes] If Yes, please specify: Syndromes Without A Name (SWAN) Australia

4. **If applicable, please specify you and/or your organisation's area of expertise.**

   We provide information and support to families/carers who have a child with an undiagnosed children or rare genetic condition. We are a peer to peer support group and advocate on behalf of our members.

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

   [Yes] Phone Number (Optional): 0404 280 441 or email: president@swanaus.com.au

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.	**Bases**: Also known as nucleotides, they are the basic components of DNA. They are denoted by the letters **A** (**A**denine), **G** (**G**uanine), **C** (**C**ytosine) and **T** (**T**hymine). The sequence of these bases forms the genetic code. **Chromosome:** A threadlike structure found in the nucleus of all the body cells (except red blood cells) consisting of DNA and proteins. Each chromosome can be thought of as a string of beads where every bead represents a gene. **Exon:** The part of the DNA message that is translated into a protein. **Mitochondria:** These structures or organelles in the cell are the main energy source and are often called the powerhouse of the cell. The mitochondria also contain their own DNA and therefore genes. Mitochondrial genes follow maternal inheritance.

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			Mitochondrial DNA: The genetic material contained in the circular genome found in mitochondria. ***Source:*** *http://www.genetics.edu.au/Publications-and-Resources/glossary-1* **Panels** Targeted gene sequencing panels are useful tools for analyzing specific mutations in a given sample. Focused panels contain a select set of genes or gene regions that have known or suspected associations with the disease or phenotype under study. Gene panels can be purchased with preselected content or custom designed to include genomic regions of interest. *Source: https://www.illumina.com/techniques/sequencing/dna-sequencing/targeted-resequencing/targeted-panels.html* *Note: Probably a better Australian definition out there*
		8. Are the definitions easy to understand? Do any definitions require amendment? If yes,	Yes, they are clear to understand

Section of Framework		Question	Response
		please provide details.	
Preamble	Pg. 7-8	9. Does the Preamble provide a sufficient overview of the Framework? If not, please provide further details.	Yes, we believe the preamble provides a sufficient overview of the Framework. We would however like to add the sentence (mostly taken from the outline of the framework): *"It is designed as an integrated Australian health system that effectively informs and supports health care for individuals and populations."* We believe the sentence would be best positioned at the end of the second "Purpose" paragraph on page 7 of the Framework.
		10. Are there linkages with other key frameworks or strategies that should be explicitly referred to in the Preamble? If yes, please provide details.	We cannot think of any other linkages with other frameworks
		11. Is a three year timeframe sufficient for the Framework? Please explain your answer.	Yes, we believe a three year timeframe is sufficient as genetics is very fluid. The fact that the scope of the Framework is intended to be flexible allows for new methods and further adaption over time.
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.	Yes, we believe they are.

Section of Framework		Question	Response
		13. Does the Strategic Context provide a clear case for improved national consistency in genomics policy? Please explain your answer.	Yes it does but we do have concerns that places like the ACT, Tasmania, NT and regional patients will be disadvantage due to location. A national approach needs to be taken, not just lead by other states competing with each other for the most funding.
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.	I think there are a number of issues, which need to be taken into consideration if we are to have an effectively integrate genomics into our healthcare system. Overall we thought the Framework addressed the issues well but as a peer to peer support group supporting many families with undiagnosed children and those of rare genetic conditions for which there is limited support, we wanted to highlight the issues that are important to us below: **<u>Equity of access</u>** • Difficult for patients who live in rural and remote areas • Diversity of populations • Difficult for CALD patients, Aboriginal or Torres Strait Islanders to access genomics • Unequable for patients with intellectual disabilities or who have mental health issues unless they have someone who can advocate on their behalf

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			• Current system sees different states having access to different services and different levels of funding • Cost prohibitive • Different levels of support depending on what hospital and what medical department you access • Age can also be a barrier **<u>Lack of resources and support</u>** • Shortage of trained geneticists • Shortage of trained genetic counsellors due to limited placements available to them • Problem that Australian genetic counsellors are not accredited and cannot provide their service through Medicare as it opens the field up to "anyone" calling themselves a genetic counsellor. If we do no have a trusted system in Australia with credibility the public will go overseas for testing and there is risk associated with interrupting results (if accurate) with very limited supports • Underfunded (or not funded at all) Peer Support groups who can follow through with patients concerns. Too often patients are forgotten once they leave medical appointments /hospitals • Research in this area and even when rare

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			conditions are discovered is extremely underfunded or non existent **<u>Resistance to change</u>** • Some doctors, more often the older generation may not be as accepting of new testing methods • Changing the general publics perceptions of personalised medicine and new testing methods **<u>Lack of knowledge</u>** • Lack of education – need a variety of mediums to educate the general public, allied health professionals and medical profession **<u>Lack of trust</u>** • Lack of confidence with new genetic testing methods • Privacy concerns, particularly around data security • What to do with incidental findings, who owns this information • Information being misused – life insurance, health insurance or given to other family members **<u>Expense</u>** • Currently genome sequencing is only available to patients who can afford it or through those

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			lucky enough to be accepted on a research basis through pilot programs such as the Melbourne Genomics Health Alliance **<u>Collaboration between health professionals and patients</u>** • Move towards more patient centred care will be met with resistance by some **<u>Lack of consistency and coordination</u>** • No rare disease policy in Australia • No formal processes by which doctors need to follow when genome testing – yes consent is given but there are no guidelines around the recommended duty of care e.g. see a genetic counsellor prior to testing and after testing results are given but there are no reference to follow up appointments. It is often when people have time to process test results that problems and questions begin to emerge. We need to be transparent and consistent in our approach. • A timely diagnosis leads to better pathways, especially for those suffering from undiagnosed and rare genetic conditions.

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A National Health Genomics Policy Framework for the next 3 years	Pg. 15	15. Are the key guiding principles appropriate? Please explain your answer.	Yes we think the guiding principles support the Framework.
Enablers	Pg. 16-17	16. Are there additional enablers that should be included? If yes, please provide details.	We think the Framework lists the main enablers, though we would like to see a National Disease Plan identified as an enabler.
Strategic Intent	Pg. 16-17	17. Is the Strategic Intent of the Framework appropriate? If no, what would you suggest?	We would like to see the word "support" added in there. "To harness the health benefits of genomic knowledge into the Australian health system in an efficient, effective and equitable way to improve **and support** individual and population health."
Priorities Areas	Pg. 17	18. Are the priority areas appropriate? Please explain why or why not.	Yes they are appropriate and we identify human resources and education as being priority areas. **<u>Human Resources</u>** • Shortage of geneticists, other medical experts who have the knowledge about genome sequencing and genetic counsellors who can support patients at intervals through their journey. **<u>Education</u>** • The general population's understanding and knowledge around genome sequencing is very

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			low and a lot of material needs to be made available in different mediums at different levels for people to comprehend the change in personalised medicine. Knowledge is empowerment for patients.
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?	Yes – we believe ethical, social and legal issues are vital if genomic medicine is to be equitable for all Australians. Community engagement with different ethical and social groups needs to be involved to gain consumer confidence.
		20. Should these issues be considered prior to the six priority areas, or after?	You cannot have one without the others. You need to build up from ethical, social and legal issues otherwise your framework won't be sustainable.
		21. Are there any other broad ethical, legal or social issues that should be addressed under this priority? If yes, please provide details.	No, we think the framework as addressed them well.
Priority Area 1 – Strong leadership and governance	Pg. 25-26	22. With regard to *Priority Area 1 – Strong leadership and governance*, is anything missing or what should change, for:	a) Current situation • Australian doesn't have a National Rare Disease Plan or Policies. • We agree with the current situations points stated.

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		(a) the current situation; (b) why is this important; (c) opportunities for improvement; and/or (d) what the future looks like?	b) Importance • Consistency and transparency amongst the States and equity amongst the States. • We agree with the other points stated. c) Opportunities for improvement • Have consumer representation involved in developing and reviewing the National Genomics and Genetics Framework. We feel the Framework lacks consumer group input from groups such as State Genetic Bodies, Rare Voices Australia and SWAN who are experts in their own fields and can represent the voice and priorities of the consumer within the Genomics Framework. • Continual bipartisan agreement and commitment to support the Framework. • We agree with the other points stated. d) The future • We agree with the draft stated points about the future but would add the work "commitment" after oversight to read "National oversight and commitment that:"
Priority Area 2 – A skilled and literate	Pg. 27-29	23. With regard to *Priority Area 2 – A skilled and literate genomics workforce*, is anything	a) Current situation • We are concerned about the lack of geneticists as currently we are led to believe there is on average an 8 month wait to see a geneticist at

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genomics workforce		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 Royal Children's Hospital in Melbourne. This impacts on families' mental health, stress levels and anxiety and needs to be addressed as a priority area. • We are concerned that consumers will go overseas and see specialists or, worse still, order genomic tests online and having no support when the results are given to the patient. (How do you even know if the tests and results are valid?) • We are also concerned with the lack of genetic counsellors and the small quota of placements in the Genetics Masters program due to limited work training placements for them. They are also not accredited and are self-regulated. Medicare does not cover their fees. • We agree with the other points stated. b) Importance • We need to utilise the experts in the fields and a multi-disciplinary approach leads to better support and outcomes for patients. • We agree with the other points stated. c) Opportunities for improvements • People working in the genomics field also need to have good communications and people skills when liaising with patients.

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			• We agree with the other points stated. d) The future • A well supported and funded genomics workforce is needed for sustainability. • We agree with the other points stated.
Priority Area 3 – Application of genomic knowledge is evidence based, high quality and safe	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 should change, for: (a) the current situation; (b) why is this important; (c) opportunities for improvement; and/or (d) what the future looks like?	a) Current situation • The general population would have no idea about the clinically validity of testing other than to trust the professionalism of their health care team. • We agree with the other current situations points stated. b) Importance • We agree with the reasons stated that it needs to become a priority. c) Opportunities for improvements • Research in Australia is underfunded but very much needed, especially when it comes to learning about rare diseases and undiagnosed conditions. We strongly support research as an opportunity for improvement along with the other points of improvements raised. • We agree with the other points stated. d) The future • We agree with the points stated about what the

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			future will look like.
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?	a) Current situation • People obtain their genomic information from a wide range of sources, but they are not always credible. People go outside Australia for genomic health. • We agree with the current situations points stated. b) Importance • Person-centred medicine gives the patient empowerment. Patients are often their own health experts. Positive health outcomes are often achieved when multi-disciplinary teams and the patients themselves are involved in treatment plans. • We agree with the other points stated. c) Opportunities for improvements • We need to focus on giving patients the opportunity to learn about genomics and precision medicine by presenting information in different mediums and suitable for a range of different ethnic and socio-economic and educational backgrounds. Clear, transparent

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			and consistent communication is going to be the key to success. Patient advocacy and peer support groups, RVA, SWAN, genomic health bodies such as the GSNV, Genetic and Rare Disease Network and groups such as the Melbourne Genomics Health Alliance Community Advisory Group would be good to engage with to ensure this is delivered smoothly. • We would like to see undiagnosed children and those with suffering from rare diseases take priority when it comes to being offered genomic testing. Once a condition is identified and understood, outcomes and disease management can lead to better treatments plans and better personalised medicine. • Some health providers may need to change their view that their expertise and knowledge is the only way. Patients are often the own health experts and respect needs to be given to them. • We agree with the other points stated. d) The future • We believe that increased genomic literacy offered in a range of mediums and versions needs to be adopted to increase the general public's understanding of genomics and precision medicine. A good understanding of

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			genomics and clear communication will be the key to consumer confidence. • Some people will be resistant to change and new methods and opportunities. • We agree with the other points stated.
Priority Area 5 – Sustainable investment in health genomics	Pg. 34-35	26. With regard to *Priority Area 5 – Sustainable investment in health genomics*, 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?	a) Current situation • Research is underfunded • We agree with the other points stated. b) Importance • Research can be cost-saving and needs to be funded and supported for advances in medicine to occur. • Governments need to stop thinking short-term and invest in consumer health as a preventative measure, it will be cost-saving in the long run. They should be proactive not reactive. • Genomic medicine has the ability to replace a lot of other medical investigations, which can be distressing to the patient and expensive and often not as conclusive. • We agree with the other points stated. c) Opportunities for improvements • Engage with consumer groups around ways they view genomic medicine becoming sustainable. d) The future • We need to leverage from other genomic

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			projects around the world and learn from their strengths to ensure Australia provides world-class genomic medicine. • We agree with the other points stated.
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?	a) Current situation • The security and integration of genomic data is a concern for consumers. • We agree with the other points stated. b) Importance • Data-sharing needs to occur not only in Australia but around the world. Often rare diseases will only be diagnosed by a match overseas due to lack of testing and the size of Australia's population. • We agree with the other points stated. c) Opportunities for improvements • Data sharing and the benefits of it need to be well communicated to patients. • Ethical guidelines will need to be developed around data and consent and should be used consistently. • The infrastructure around data sharing needs to be further developed, maintained and funded. • We agree with the other points stated. d) The future • Secure data collection and data sharing

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			processes will be fully funded by Government. • We agree with the other points stated.
Implementing the Framework	Pg. 39	28. Is the suggested approach to implementing the Framework reasonable and appropriate? Please explain your answer.	Yes, we like the idea of having a governance body that is responsible for developing the genomics action plan. It would be great if it could include a cross-section of people, not just policy makers and health professionals. There are active health consumer groups such as RVA, genetic bodies and SWAN who would have an active interest in genomic medicine.
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.	Yes, we thought the Framework was well thought-out, clear and easy to follow. The section headings and dot points made it easy to follow.
		30. How could the review and evaluation of the Framework be strengthened?	By having more consumer/patient advocacy and peer support groups involved in the process. They are the ones dealing first hand with the health care system.
		31. Do you have any other feedback on the Framework?	Thank you for the opportunity to comment and is great that the Government has put a lot of time and thought into the draft National Health Genomics Policy Framework. However, we were a little disappointed more consumer groups and peer support were not personally invited to the consultation or sent a draft copy of the Framework for comment. I don't believe

Section of Framework		Question	Response
			enough consumer/patient advocacy and peer support groups knew about the forums or were aware of the opportunity to give feedback on the National Health Genomics Policy Framework.
		32. Are there any issues you would like covered at the stakeholder consultation forums in February 2017?	Thank you we found the comments and discussions useful around the Framework.
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?	Yes I would like the outline to encompass the word Genetics as well. “Outline of the National Health Genomics **and Genetics** Framework” and the Health System Policy Intent to read “An integrated Australian health system that effectively informs **and supports** health care for individuals”.