

Psychosocial considerations when offering genetic testing in the ALS population

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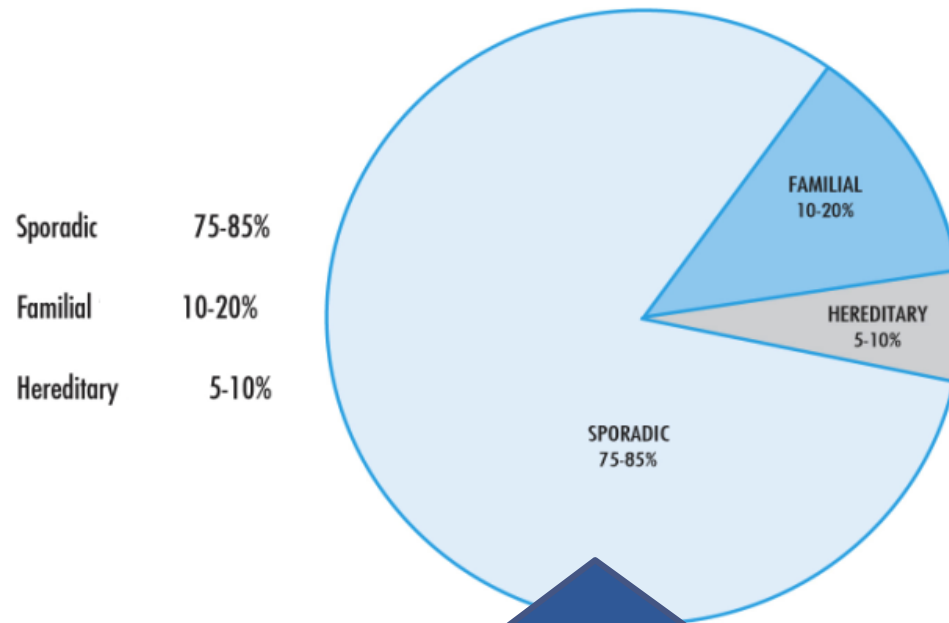
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Causes of common disease

Distribution of Sporadic, Familial, and Hereditary



~7-10% of isolated ALS
have gene mutations
(C9orf72 + SOD1)
equally found in fALS

What genes are we testing?

Based on personal and family history

fALS, including FTD

sALS
Any age

Early onset ALS

Comprehensive panel:
C9orf72 repeat expansion
Multi-gene panel NGS
(SOD1, TDP, FUS)
Deletion/duplication

Limited panel:
C9orf72 repeat expansion
SOD1 sequencing

Genetic testing for affected patients

- Pre-test counselling provided for all newly dx ALS
- Discussion:
 - Pedigree – for risk assessment
 - Explanation on cause of disease
 - Basic genetics - genes, mutation, autosomal dominant transmission
 - Indication for why we are offering genetic testing
 - Possible results
 - Mutation identified
 - Mutation not identified
 - VUS (Variant of unknown significance)
 - Review implications for patient and family members
 - Discuss motivations

Genetic testing for affected patients

Why patients are accepting or refusing genetic testing

Accepting

- Potential treatment
- To inform children of risk
- Understand why they have the disease
- “Because my doctor wants me to”

Refusing

- To NOT inform children of risk

Conflict & Struggles

- Patients struggling between wanting to know their genetics for potential treatment vs not wanting to tell their children
- Isolated cases: were reassured no one else in family affected

Some patients:

- see only the positive in doing this testing
- see the fear in having to inform family members
- are so hopeful for a treatment, they will deal with family issues later

Our experience in clinic

	Total	fALS	sALS
Total tested	170	17	147
# of mutations identified	24	14	10
C9orf72	13	6	7
SOD1	9	8	1
other	2	0	2
	24/170= 14%	14/17 = 82%	10/147= 7%
* 7 Patients have refused			

Only when a gene is identified in a family, can we offer gene testing to unaffected family members

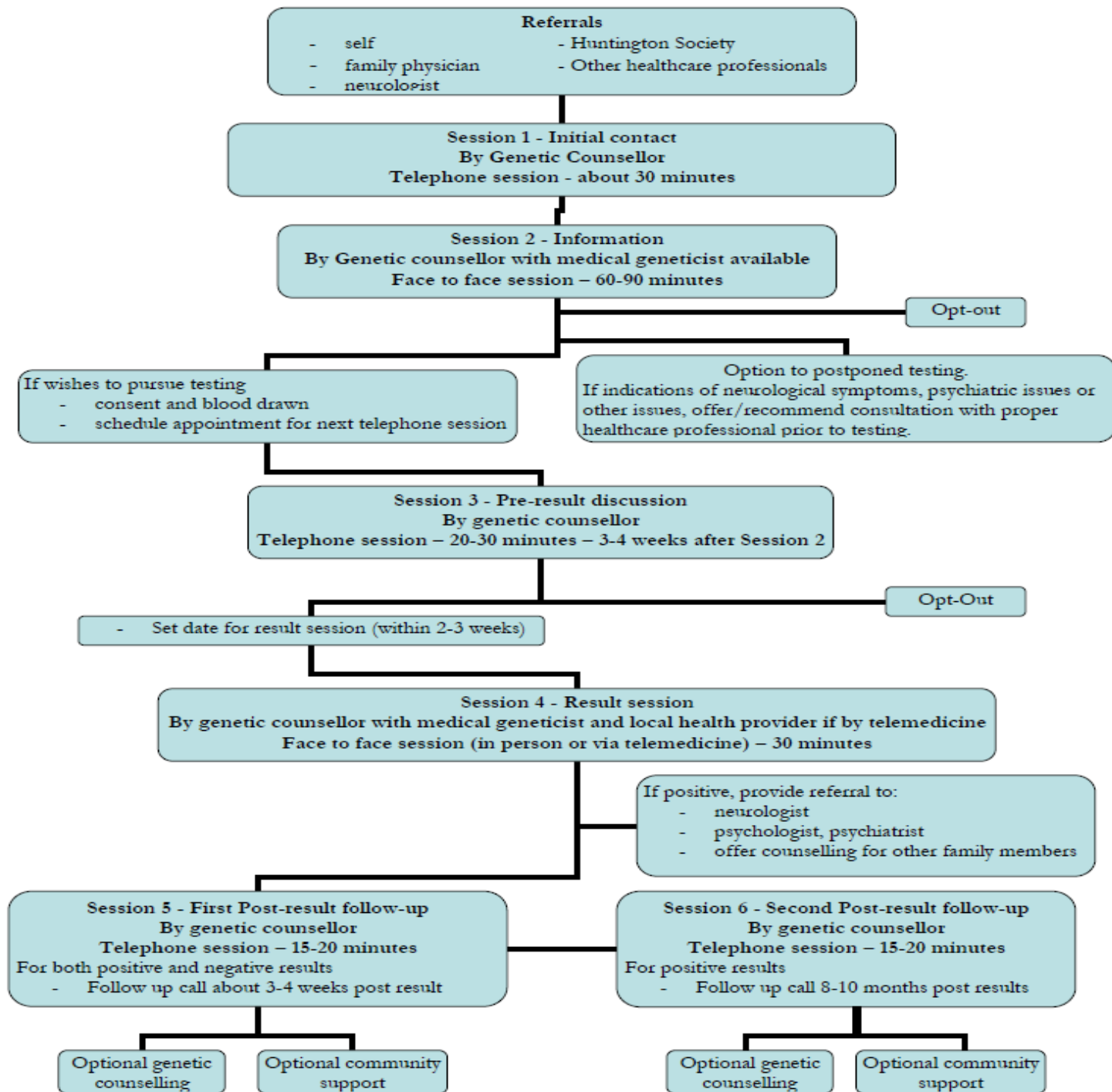
Predictive testing in asymptomatic members

- $\frac{1}{2}$ chance of carrying the family gene mutation or not
- Cannot predict age of onset or progression of disease



Predictive protocol

- Follow the Huntington Disease (HD) predictive protocol for all neurodegenerative diseases
 - Neurologist, genetics, psychiatrist/psychologist
 - Self-selective
 - What HD studies show:
 - 2 months following results are most turbulent
 - Post-test emotional state 1 yr after results, return to pre-test emotions
 - Long-term support is necessary for + carriers – value in multidisciplinary clinics
 - The percentage of test usage was lower in untreatable diseases.
 - Only 2 published articles specific for ALS



Psychosocial implications in predictive testing

- Reasons for testing can vary according to:
 - Stage in life they are in (different considerations in your 20s, 30s, 40s, 50s, retirement years)
 - How many more years left before the disease will manifest
 - Experience with disease
 - Reduce uncertainty
- Discussion
 - What will change in their life, should they be positive or negative?
 - What changes will they make, should they be positive or negative?
 - Who / what is their support group like?
 - What is their "normal" emotional state like and what are their "go-to" tools to help themselves
- Views and impact

Many people from the outside think it's so devastating....

 - But what they are really doing is choosing their values, what is important for them given the time left
 - Much emotional difficulties to process;
 - Grief of, not having children, losing a partner
 - Fear of living through the disease alone,
 - Questions / choices ... how will it be to have the disease / career choice

Change is difficult to make

 - Do you really need a genetic test to create this change?
 - Emotions will evolve as time progresses

Goal of doing predictive testing is to create positive change in their remaining healthy lives

Thank you!

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